



# The *Gathered View*

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

## An International PWS Conference—Italian Style *IPWSO Meeting Coming to America in 2001!*

Opera, sumptuous meals, the canals of Venice, the beaches of the Adriatic, and, of course, the warmth of the Italian host association made this spring's International Prader-Willi Syndrome Organisation (IPWSO) conference a unique and rich experience.

Some 22 parents and professionals from the United States joined several hundred of their counterparts from 23 countries to participate in the Third Prader-Willi Syndrome International Scientific Workshop and Conference May 21-24 in Lido di Jesolo (Venice), Italy. PWSA's delegation included Executive Director Janalee Heinemann, Scientific Advisory Board members Suzanne Cassidy, Jeanne Hanchett, Barbara Whitman, and Rob Nicholls, Board Member Fran Moss and former board member Louise Greenswag, and Parent Representative to IPWSO Mildred Lacy.

Mildred, who was elected IPWSO vice president at the meeting, reports that growth hormone was the "hot topic" of the week's presentations and that the Saturday evening gala was the social highlight for all:

"Our president-elect, Giorgio Fornasier, just happened to be a professional opera singer, and he performed several operatic renditions along with secretary-elect Monika Fuhrmann, also a professional opera singer. Oh, what joy they brought to everyone that attended!

"All the individuals with Prader-Willi syndrome (25 participants) honored us

with a parade, dressed in their finery. The custom is to form a line of dancers with music, and everyone there is to join in, and I must say it was quite a line that snaked around that beautiful hall! This was to signify a warm welcome to all participants."

### **IPWSO Business**

For those of us in the United States, the biggest news from this year's IPWSO conference is that PWSA (USA) won the bid to host the 4th International PWS conference in the year 2001. Thanks to Joan and Jim Gardner, who created an excellent display and presentation for the IPWSO board to consider, the next IPWSO conference will likely be held in Minnesota, the birthplace of our national association.

Other IPWSO news:

- Tiina Silvast of Finland won the contest to design a logo for the international association and will receive free registration to a future IPWSO conference.
- Kumiko Egawa from Japan won the poster award for her display describing

the oral feeding of an infant with PWS. Hers was one of 29 posters from 15 countries on a range of topics. *(Continued on page 3)*



**Mario Bianco, a young member of the Associazione Italiana PWS, offers a welcoming smile to PWSA (USA) members attending the IPWSO Conference in May.**

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## Executive Director's View

by Janalee Heinemann

Until May, Italy, Kenya, Malaysia, Australia, Japan, and New Zealand were just all places on a map to me. Then, in attending the IPWSO international conference in Lido de Jesolo, Italy, they all became very real through the parent and professional attendees. The conference was alive with conversation, color, and warmth, as we all eagerly shared and learned. This was not a conference where people were there to compete. Too little is known on both a medical and psychosocial level to feel a need to outdo each other or guard our resources. Everyone was eager to give and to get the essential threads of knowledge that will eventually lead to weaving a better quality fabric of life for all of our children with Prader-Willi syndrome.

I was touched by many special moments during the conference. First, by our special Italian hosts who put on a wonderful conference in spite of only having a year to prepare. One of the special events that I'm sure will stand out in everyone's mind is the gala evening, with our new IPWSO president, Giorgio Fornasier, treating us to his outstanding opera singing. There was course after course of fabulous Italian food—including a flaming entrée—wine flowing, and delightful company. Two scenes that evening especially touched my heart. One was when all of the conference participants, young and old, parents, children, and professionals, spontaneously created a long chain and danced around the room. They were the living proof of the international fabric of PWS. The other was when a little boy with PWS was running across the room holding his father's hand. I looked across the table and saw my husband, Al, and Paul Paolini (Calif.) both looking at that child with the same look of sheer love and delight written across their faces. Paul's son is deceased, and our son is grown and out of the home, but the fabric that bonds us together comes with a lifetime guarantee.

I came away from the meeting with a greater respect for all of the countries represented. I think sometimes in America we tend to think we are more advanced in everything. But, at the conference, I came to appreciate that some countries are more advanced in caring for their families with special needs, and even those countries with no services or awareness regarding PWS have educated, courageous parents who will be the pioneers for a better future. There were no miraculous breakthroughs announced and no magic pill unveiled, but I think we all came away with a feeling that with 24 countries represented and 300 people in attendance, together we will weave a beautiful tapestry of knowledge, courage, support, compassion, and understanding.

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Opinions expressed in *The Gathered View* are those of the authors or editors and do not necessarily reflect the views of the officers and board of directors of PWSA (USA). *The Gathered View* welcomes articles, letters, personal stories and photographs, and news of interest to those concerned with Prader-Willi syndrome.

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- Retiring president and founder of IPWSO Jean Phillips-Martinsson of England was presented a certificate signed by board members and officers and given an engraved gold medal.
- IPWSO is now a member of the European Disability Forum, which will afford the organization greater credibility in seeking foundation grants.
- There are now 24 member countries in the IPWSO, three of which are associate members with no PWS chapters. IPWSO officials welcomed two new member countries: Austria and Israel.
- Elections were held for IPWSO officer positions, scientific board, and editor positions. The following members were elected for a three-year term:

President—**Giorgio Fornasier** (Italy)

Vice-President—**Mildred Lacy** (USA)

Secretary—**Monika Fuhrman** (Germany)

Treasurer—**David Gordon** (South Africa)

Scientific Board—**Martin Ritzen, M.D.** (Sweden) and **Ellie Smith, M.D.** (Australia)

Editor, parent newsletter—**Cindy Adams-Vining** (New Zealand)

Editor, scientific newsletter—**Susanne Blichfeldt, M.D.** (Denmark)

Dr. Suzanne Cassidy resigned from the IPWSO Scientific Board but agreed to be available as a consultant to the board.

**Giorgio Fornasier, new president of IPWSO, shares a laugh with retiring president and founder Jean Phillips-Martinsson during the Gala Evening activities at the IPWSO conference.**



❖  
*Editor's note:* Summaries of some of the IPWSO conference presentations will appear in our next issue of *The Gathered View*, along with reports from the PWSA scientific conference in July.

## Iowa Joins States With Laws Naming PWS

by Tammy Davis  
President, PWSA of Iowa

Iowa has added PWS into the developmental disability code! We approached the House of Representatives of Iowa on March 23 of this year, spoke with a few members of the human resource sub-committee about the challenges of raising a child with PWS and the challenges of placing an adult with the syndrome. There was a bill open at the time we met, and House of Representatives people and the man who heads the Department of Human Services for Iowa felt we could add it to this bill and get it through this session. It was written into House File # 2558 under Mental Health, Developmental Disabilities, Section 16, ACCREDITATION OR CERTIFICATION OF SERVICE PROVIDERS, and reads:

*“Effective July 1, 1998, the department of human services shall include persons with Prader-Willi Syndrome who, due to their disability, experience limitations of one or more of the major life activities as defined in the Developmental Disability Assistance and Bill of Rights. Pub. L. No. 101-496, in the definition of ‘persons with developmental disabilities’ used in the department’s accreditation or certification of providers of services for persons with mental illness, mental retardation, and developmental disabilities.”*

It passed and was signed by the governor.

A lot of words for a very simple task!!! (But that is our government!) And, although it’s now in Iowa law, it guarantees NOTHING!!!! It is a foot in the door, so to speak. An admission that this syndrome does exist and is life threatening. Every county in Iowa disperses the funding money in different ways; they are not obligated by any law to have funds available for any specific category of developmental disability. We as parents must find our central point of coordination (CPC) in our county and make sure there are funds for the type of services we need. The money is there—how they spend it is another story.

My advice to parents in Iowa is to access the system early in life—it will make a difference!

*Editor’s note:* At least four state codes now have PWS specifically named as a disability eligible for services—those in Connecticut, Florida, Wisconsin, and now Iowa.

# Growth Hormone Therapy: Tackling the Funding Issue

by James and Carolyn Loker  
Co-Presidents, PWSA of Michigan

In last November's *Gathered View*, we were given preliminary information on the impressive improvement of individuals with Prader-Willi syndrome on growth hormone replacement therapy. This improvement included not only increases in height but also a reduction in total body fat. We had our daughter tested when she was 2½ years old, which showed her to be growth hormone deficient. When we decided to start her on growth hormone, our insurance company initially refused to cover it, saying that growth hormone replacement is considered experimental in Prader-Willi syndrome. While appealing this decision, we started Anna on growth hormone through a program set up by the drug company. Eventually we were able to convince the insurance company that, since she had growth hormone deficiency, they could not discriminate due to the presence of Prader-Willi syndrome.

Since she has been on growth hormone, in six months she grew three inches (from the 5th percentile to the 30th percentile); she had a weight gain of 2 lbs. (staying at the 25th percentile); but more importantly, her body fat index decreased from 60 percent to 40 percent, and her arm muscle area increased from the 25th to the 65th percentile. In addition, she is more active, has improved muscle tone and balance, and is verbalizing more. Interestingly, we also noted a dramatic increase in the size of her hands and feet, with her shoe size increasing from a 5 to an 8.

**If you are interested in starting your child on growth hormone, here is a checklist of what to do:**

1) *Work with your local physician* to locate a pediatric endocrinologist who is willing to test your child for growth hormone deficiency. This usually entails drawing blood tests and giving your child some medications that stimulate the pro-

duction of growth hormone. Levels less than 10 ng/ml indicate growth hormone deficiency. They will also obtain X-rays of your child's hand to determine bone age as well as plotting their height, weight, and other measurements. You may have some difficulty convincing them to test your child depending on age, etc. Be prepared with information and articles detailing the benefit of growth hormone in individuals with Prader-Willi syndrome.

2) *Decide with your endocrinologist which drug company to work with* (since they all are using recombinant human growth hormone, there is not much difference in the drugs). Contact your insurance company, as they may have a list of companies they work with. There is no reason to delay starting treatment while waiting for the insurance company, as all three manufacturers will supply growth hormone during the process of finding coverage. Have your endocrinologist contact the drug company's program director (numbers listed at right) to get started on growth hormone treatment. A case manager from the drug company will contact you to get the necessary information to process a claim with your insurance.

3) *If your insurance company does not approve, you have the right to appeal* the decision. The case manager will help guide you in the appeal process. It may be helpful to include information on the benefits of growth hormone to the insurance company during the appeal. During the appeal, growth hormone may be provided by the drug company.

4) *If the insurance company still refuses to fund it, or if you do not have insurance, work with the drug company* to find alternate sources to help pay for it. This may include children's special health care services. All three companies also have a Support Program that will supply growth hormone at reduced costs depending on your income, number of dependents, and family assets.

## ***GH Insurance Problems Noted In Recent Study***

A study published in the March issue of the *Journal of the American Medical Association* found that insurance coverage for growth hormone treatment varies widely among insurers and in many cases does not support the doctors' recommendations. Researchers from Case Western Reserve University's Department of Pediatrics compared insurance coverage decisions regarding growth hormone therapy for children with growth hormone deficiency, Turner syndrome (the only syndrome for which the FDA has specifically authorized its use), or renal failure. Although GH was recommended by the child's physician in 78 percent of cases studied, 28 percent of these were denied coverage by their insurers. For Turner syndrome alone, the results were even more dramatic: GH treatment was recommended by the physician in 96 percent of cases, but insurers provided the GH coverage for only 52 percent of those patients.

Finkelstein et al., 1998. Insurance coverage, physician recommendations, and access to emerging treatments: growth hormone therapy for childhood short stature. *JAMA* 279(9), p. 663-668.

## **Growth Hormone Manufacturers:**

**Genentech —1-800-545-0498**  
(Products: Nutropin, Protropin)  
[www.gene.com/](http://www.gene.com/)

**Eli Lilly —1-800-847-6988**  
(Product: Humatrope)  
[www.lilly.com/](http://www.lilly.com/)

**Pharmacia & Upjohn**  
**1-800-645-1280**  
(Product: Genotropin)  
[www.genotropin.com/](http://www.genotropin.com/)

**Other sources of support  
and information on funding  
for growth hormone therapy:**

**The Magic Foundation**  
1-800-3MAGIC3  
[www.magicfoundation.org/](http://www.magicfoundation.org/)

**Human Growth Foundation**  
1-800-451-6434  
[www.genetic.org/hgf/index.shtml](http://www.genetic.org/hgf/index.shtml)

## Vanderbilt University PWS Program Project Request for Subject Participation

Merlin G. Butler, M.D., Ph.D., and Travis Thompson, Ph.D.  
Vanderbilt University and The John F. Kennedy Center, Nashville, TN

Researchers at Vanderbilt University are in the midst of a unique, comprehensive study of weight, exercise, and health in people with Prader-Willi syndrome (PWS). Dr. Merlin G. Butler, Director of the Regional Genetics Program in the Department of Pediatrics, and Dr. Travis Thompson, Director of Vanderbilt University's John F. Kennedy Center, head a team of 20 scientists and specialists from 12 disciplines. They have received funds from the National Institute of Child Health and Human Development to study 50 people with PWS and to compare with 50 people of similar ages, learning ability and body weight, who do not have PWS. To date, we have enrolled 57 subjects (PWS and control) in our study since beginning in May 1995.

The purpose of this study is to discover the steps along the pathway from the genetic information to behavior of a person with PWS. We hope to better understand the cause of Prader-Willi syndrome and to treat people with this condition more effectively. In order to do so, our subjects complete a series of assessments at Vanderbilt University which requires about two days on each of two separate visits. An eight-week exercise program is scheduled between the two visits in order to determine changes, if any, in metabolism from exercise. The assessments are not painful and are relatively noninvasive. A fitness test on a treadmill will be performed; blood will be drawn for genetic and metabolic studies; a vision assessment and eye examination will be completed; a food preference test will be administered; and an MRI (to determine body composition and percent body fat and possibly brain function) and a bone density scan (to determine bone mineral content) will be completed. On both visits, the participants will spend eight hours in a metabolic room which determines how many calories are burned during normal and exercise activities.

Who is eligible to participate in this study? We are currently looking both for people with PWS and for obese controls with mild to moderate mental retardation. We are specifically interested in finding PWS subjects with maternal disomy of chromosome 15 (both 15s from the mother) and PWS subjects with atypical or unusual genetic findings such as translocations involving chromosome 15, imprinting mutations, or smaller chromosome 15 deletions. We hope to study equal numbers of PWS subjects with the deletion of chromosome 15 and with maternal disomy 15 (seen only in 25 percent of PWS subjects); therefore, we need more PWS subjects with maternal disomy.

Medical conditions and medications will be reviewed by a physician to determine whether a person would be a good candidate for the study. In return, the participants will receive at no charge, a comprehensive medical assessment, psychological and educational evaluations, and subject participation money and reimbursement for mileage. Lodging and meals for the study participants and their parents will be provided. Specific information gained from the various assessments will be provided to the parents in a report following completion of the assessments. It is hoped that these reports will help the participant's family and doctor in treating their special needs.

This study is an important step in our search for solutions to the many unique problems seen in persons with Prader-Willi syndrome. Our study may also shed light on the cause and treatment of obesity in the general population, as well as eating disorders such as bulimia or anorexia nervosa.

If you are interested in further information regarding participation in our research project, please contact Elizabeth Roof, Project Coordinator, at 615-322-8982.

Research announcements are published as a courtesy to professionals conducting research studies on Prader-Willi syndrome. Unless otherwise noted, they have not been reviewed or sponsored by PWSA (USA). Interested members should contact the researchers directly for information.

### New Articles on PWS

- Butler MG, et al. (1998) Photoanthropometric study of craniofacial traits in individuals with Prader-Willi syndrome on short-term growth hormone therapy. *Clin Genet*, April 1998, 53(4): 268-275.
- Davies PS, et al. (1998) Effect of growth hormone on height, weight, and body composition in Prader-Willi syndrome. *Arch Dis Child*, May 1998, 78(5): 474-476.
- Dearlove OR, et al. (1998) Anaesthesia and Prader-Willi syndrome, 8(3): 267-271.
- Eiholzer U, et al. (1998) Treatment with human growth hormone in patients with Prader-Labhart-Willi syndrome reduces body fat and increases muscle mass and physical performance. *Eur J Pediatr*, May 1998, 157(5): 368-377.
- Hart PS. (1998) Salivary abnormalities in Prader-Willi syndrome. *Ann NY Acad Sci*, April 1998, 842: 125-131.
- Kamel A, et al. (1998) Effect of growth hormone treatment on insulin action in adipocytes from children with Prader-Willi syndrome. *Eur J Endocrinol*, May 1998, 138(5): 510-516.
- Pietrobelli A, et al. (1998) Prader-Willi syndrome: relationship of adiposity to plasma leptin levels. *Obes Res*, May 1998, 6(3): 196-201.
- Sills IN, et al. (1998) Non-insulin dependent diabetes mellitus in a prepubertal child with Prader-Willi syndrome. *J Pediatr Endocrinol Metab*, March 1998, 11(2): 281-282.
- Smith IE, et al. (1998) Treatment of ventilatory failure in the Prader-Willi syndrome. *Eur Respir J*, May 1998, 11(5): 1150-1152.
- van Lieshout CF, et al. (1998) Problem behaviors and personality of children and adolescents with Prader-Willi syndrome. *J Pediatr Psychol*, April 1998, 23(2): 111-120.

### CORRECTION

The last issue of *The Gathered View* mentioned a 1997 overview article by Dr. Suzanne Cassidy that appeared in a British journal under the heading "Syndrome of the Month." The title of this seven-page article is simply "Prader-Willi Syndrome," and the correct name of the journal in which it was published is the *Journal of Medical Genetics* (Nov. 1997, Vol. 34, No. 11, pp. 917-923).

# Recent Advances in the Research and Treatment of Obesity: How They Relate to the Prader-Willi Syndrome

by Daniel J. Driscoll, Ph.D., M.D., and Helen McCune, M.S., R.D.  
Pediatric Genetics, University of Florida College of Medicine, Gainesville, Florida 32610

*Dr. Driscoll spoke on obesity research at the 1997 PWSA National Conference in Orlando and promised to summarize this important information in an article for The Gathered View. He and dietitian Helen McCune collaborated to develop the following piece, which offers practical advice on weight management, as well as giving an overview of the latest obesity research and recent developments in medications for weight control.*

## Background

Recently there have been several discoveries that have underscored the important role genetics plays in obesity. In this article we discuss obesity in general, several mouse genetic models and human genetic obesity syndromes (particularly the Prader-Willi syndrome), and finish with various treatment strategies for obesity, especially those relevant to the Prader-Willi syndrome (PWS).

The definition of obesity in adults (i.e., those individuals over 18 years old) is typically based on a body mass index (BMI) of greater than 27, with morbid obesity being defined as BMI greater than 40. The BMI is calculated by taking weight in kilograms and dividing it by height in meters, squared ( $\text{kg}/\text{m}^2$ ). [See chart, page 9.] The determination of whether a child is overweight is done by comparing their weight to an ideal body weight (IBW) calculated from standard growth curves. A child with a weight exceeding 120 percent of the IBW is considered obese.

At least 25 to 30 percent of adult Americans are considered overweight. Obesity is a risk factor in five out of the top 10 causes of death in this country (heart disease, stroke, diabetes, atherosclerosis, and various cancers). The prevalence has been increasing, particularly in children. Among those with PWS, it is the rare individual who has not been obese at some point in their life, and for all individuals with PWS weight control is a constant key management issue.

## Mouse Models

In December of 1994, Jeff Friedman and colleagues at the Rockefeller Institute in New York made a landmark breakthrough in obesity research. They

discovered the genetic defect in the *obese* (*OB*) mouse. The *obese* mouse had been a genetic model for several decades, but it was not until Dr. Friedman's discovery that the genetic defect was found. This finding has led to a series of discoveries by other investigators, which has resulted in a better (but by no means complete) understanding of the cascade of events leading to obesity.

The *obese* gene makes the protein leptin (Greek for "seeking thinness"). The leptin protein is a hormone made in the fat cells which feeds back to the hypothalamus in the brain. The body's response to increased leptin is to decrease eating, increase energy use, and increase sympathetic (i.e., a catabolic, energy expending system) activity. When leptin levels are low, the hypothalamus sends "signals" to increase food intake, decrease energy output, decrease reproductive function, decrease temperature, and increase parasympathetic (i.e., an anabolic, energy conserving system) activity.

However, several research groups have shown that leptin levels are not low in the PWS population, and in fact the greater the obesity the higher the leptin levels. This finding is identical to obesity in the general population, suggesting that most causes of obesity in humans, including in PWS, are due to central problems in the brain, probably in relationship with the hypothalamus in the brain.

There are at least five very good naturally occurring mouse genetic models for obesity which include those named *obese*, *diabetes* (which has a deficient leptin receptor in the hypothalamus), *fat*, *tubby*, and *agouti*. These mouse models have enabled scientists to discover and study various neurotransmitters in the brain that are important in appetite regulation and body mass.

Neurotransmitters made in the hypothalamus in the brain that are noted to play an important role in obesity include neuropeptide Y (NPY) and galanin. High levels of NPY increase body weight, particularly by increasing carbohydrate intake, and decrease learning and memory. High levels of galanin will increase fat intake and impair learning and memory. High levels of two other proteins (UCP1 and UCP2) are known to increase energy use.

Recently three different groups (led by Dr. Bruce Cattanach in England, Dr. Cami Brannan at the University of Florida, and Dr. Rob Nicholls at Case Western Reserve University) have created mouse models for PWS. Unfortunately these mice all die at a young age due to "failure-to-thrive." Once investigators learn how to enable the mice to survive this early phase, these mice will become invaluable tools in our increased understanding of PWS and facilitate therapeutic trials.

## Human Genetic Obesity Syndromes

There are several genetic syndromes besides PWS which have obesity as a cardinal component. These include the Cohen, Bardet-Biedl, Albright Hereditary Osteodystrophy, Borjeson-Forssman-Lehmann, and Carpenter syndromes. Of these conditions, PWS is the best known and has been the most extensively studied at the clinical and molecular level. Interestingly, individuals with PWS are initially very hypotonic and labeled as "failure-to-thrive" as neonates. All babies with PWS need some sort of assistance in feeding in the first several months of life. Usually this takes the form of nasal gastric or oral gastric tube feeding. The diagnosis is frequently not made until 2 to 5 years of age, at

which time these patients start to accelerate off their weight curves. At this age they typically become obese and have hyperphagia (i.e., very aggressive food-seeking behavior).

In 1981, Dr. David Ledbetter, then at Baylor University, showed that many of the Prader-Willi patients had a small deletion in chromosome 15 at bands q11 to q13. We now know that PWS is an example of genomic imprinting. Imprinting is where a particular gene is expressed differently in an individual depending upon whether it came from the mother or the father. Only a few of the estimated 100,000 genes in the human genome are imprinted.

All individuals with PWS have a deficiency of paternal (from the father) inheritance in 15q11-q13. There are three main molecular classes of Prader-Willi patients: approximately 70 percent are the result of a paternal deletion (3-4 million base pairs of DNA) in proximal 15q; 25 percent are due to maternal uniparental disomy, or UPD (two chromosome 15s from the mother and none from the father); and the third class of patients (about 5 percent) are termed imprinting mutations (a very small deletion in the center that sets the "imprint" for the 3 to 4 million base pair region of 15q11-q13.) An opposite, and very different, syndrome occurs when there is a maternal deficiency of the same region. This syndrome is the Angelman syndrome, in which affected individuals typically do not have obesity but do have severe mental retardation, a severe speech impairment, a movement disorder characterized by an unsteady gait and upraised hand movements, and a very distinctive neurobehavior with an inappropriately happy affect.

The Prader-Willi syndrome is felt to be due to a deficiency of *several* paternally inherited genes. The father's chromosome is typically normal, but the defect arises by one of the three mechanisms listed above during the father's spermatogenesis (and the mother's oogenesis for the UPD cases). Therefore the Prader-Willi syndrome fits into the class of genetic syndromes called "contiguous gene syndromes" (i.e., the loss of several genes leading to the collection of findings in a particular syndrome). In chromosome 15q11-q13 several genes are known to be only expressed (functional) when they come from the father. These genes include *SNRPN*, *ZNF127*, *NECDIN* and *IPW*. There are probably several other genes

in this region which remain to be characterized that will also only be expressed when inherited from the father, and these genes will all be candidates to be involved in the pathogenesis (i.e., clinical features) of PWS.

The genes listed above are now being investigated to determine their role in PWS. One strategy is to examine patients with many, but not all, the features of PWS. These patients, whom we have termed "PWS-like," have obesity and learning problems, as well as many of the neurobehavioral characteristics of PWS. These patients do not typically have the neonatal failure-to-thrive or the characteristic Prader-Willi face. Perhaps the PWS-like patients only have a mutation in one of the imprinted genes in the PWS region. Understanding the mechanisms involved in the pathogenesis of PWS and the PWS-like conditions will give invaluable insights into the causes

**Understanding the mechanisms involved in the pathogenesis of PWS and the PWS-like conditions will give invaluable insights into the causes of obesity and clues as to future rational treatment strategies.**

of obesity and clues as to future rational treatment strategies.

### **Treatment Strategies**

Sometime after 1 year of age, the individual with PWS typically starts to develop hyperphagia and become obese. If left untreated, this can have devastating consequences: morbid obesity leading to a significantly increased risk of developing congestive heart failure, hypertension, diabetes, obstructive sleep apnea, and premature death.

Therefore, a crucial component of caring for the individual with PWS is avoiding or treating obesity. This involves reducing energy (calorie) intake, increasing physical exercise, behavioral and environmental management, and possibly various pharmacological (drug) treatments. The ideal treatment goal in adults, is to achieve a body mass index (BMI) of 21-26. However, even if a BMI of 21-26 cannot be reached, a 5- to 10-percent reduction in body weight can achieve health benefits such as a de-

crease in blood pressure and blood glucose. In children, the goal is to achieve a body weight of 100-120 percent of ideal body weight (IBW).

### **Lifestyle Modifications**

Various types of **diets** such as ketogenic, hypocaloric protein-sparing, and low-sugar have been used in the treatment of obesity associated with PWS, but there have been no studies which indicate that one type of low-calorie diet is most beneficial. However, there is some evidence, with normal obese individuals, that a low-fat diet may enhance weight loss.

Selection of a weight loss diet should meet the individual's nutrient requirements and be acceptable to the individual and his or her family or care givers. Since the energy requirements of individuals with PWS are lower than normal individuals' by as much as one-third to three-fourths, the calories needed to lose weight are very low — 7-8 kilocalories per centimeter of height per day or 17-20 kcal/inch/day, which translates to 1,000 to 1,200 calories a day for adults and 600-800 calories for children. A low-fat diet (25 percent of total calories) that provides the Recommended Dietary Allowance (RDA) for protein should be encouraged because it appears to enhance weight loss and permits a greater quantity of food. It is difficult to meet an individual's vitamin and

mineral requirement on a low-calorie diet, so a vitamin and mineral supplement is recommended. Calcium intake should be carefully monitored to ensure adequate intake because individuals with PWS are susceptible to osteoporosis. Often an additional calcium supplement is necessary. Finally, very low-fat diets should be assessed for essential fatty acid adequacy, and if low, the diet should be supplemented with essential fatty acids.

**Exercise** should be encouraged daily. Not only does it increase caloric expenditure, but in normal individuals, exercise increases the desire for high-carbohydrate food instead of high-fat foods. In studies involving normal obese subjects, high-intensity exercises followed by low-intensity exercise appears to be more effective at promoting weight loss than low- to medium-intensity exercise that involves the same number of expended calories. A treadmill works well because it can be programmed for more intense exercises.

(Continued from page 7)

Lastly, **behavioral and environmental management** is essential for successful weight loss. Individuals with PWS typically achieve their greatest success and sense of well-being when involved with a highly structured system. Limiting food accessibility is crucial to this success (frequently this means locking food cabinets and refrigerators). In addition, structured meal times should be established. There has been a lot of debate about the use of food as reinforcer. In the Alachua County ARC group homes in Gainesville, Florida, they have developed individual programs where food is used as an incentive for exercise, and as a reward if certain behaviors are maintained. This program has been very successful in promoting daily exercise and weight loss.

### Pharmacotherapy

There are several classes of drugs that have been used with varying success in treating obese patients in general. The various classes of drugs include appetite suppressants, thermogenic agents, and digestive inhibitors. Each has its strengths, weaknesses, and side effects. Medications for treating obesity, particularly in the PWS population, should be used cautiously and be prescribed only by physicians knowledgeable about treating obesity and PWS. In addition, medications should be used only in conjunction with the active involvement of a nutritionist to develop a dietary plan. Finally, it must be emphasized that each person is an individual, and some medications work better for some than others. There is not enough time to discuss all the medications used in obesity, and therefore what follows is a discussion of a select few relevant to PWS from each of the main classes.

Several different medications with different modes of action have been used in the **appetite suppressant category**. Medications like phentermine (e.g., Adipex, Fastin, Ionamin, and Zantryl) are **centrally acting adrenergic agents**. Phentermine has an effect similar to amphetamines but has a decreased risk of abuse. The most common side effects are secondary to its stimulant effects. The **serotonin-reuptake inhibitors** have also been shown to reduce appetite, probably due to serotonin's effect on the brain in making an individual feel less



hungry. Various drugs that belong to this group include fenfluramine (Pondimin), dexfenfluramine (Redux), fluoxetine (Prozac), and fluvoxamine (Luvox). Recently sibutramine (Meridia) has been approved by the Food and Drug Administration (FDA) for the treatment of obesity. This medication is a combination of both an adrenergic and serotonergic agent. It has only recently been released, so its long-term efficacy and potential serious side effects are largely unknown, particularly in the PWS population. We have recently used it in two individuals with PWS with good effect.

In September 1997, the FDA requested the voluntary withdrawal of dexfenfluramine/Redux (which had only been approved by the FDA for the treatment of obesity a year earlier, after many years of use in Europe without incidence) and fenfluramine/Pondimin (half of the widely used and very successful fenfluramine-phentermine, or "fen-phen," drug combination) due to an increased incidence of valvular heart disease in patients taking them, particularly in the fen-phen combination. Phentermine alone, however, is still available for use, but its overall efficacy is reduced in appetite suppression compared with the combination of the two drugs with different actions. The withdrawal of these drugs from the market has made everyone more cautious about prescribing medications for the treatment of obesity.

The **thermogenic agents** (such as the combination of ephedrine and caffeine) work by increasing energy expenditure thereby resulting in weight loss. They have not been extensively used in the PWS population, and it is therefore difficult to predict their efficacy.

The final group of drugs for the treatment of obesity are the **digestive inhibitors**. Representative of this group is orlistat (Xenical), which is currently awaiting final FDA approval. These drugs work by interfering with the breakdown and digestion of dietary fat, leading to malabsorption. A serious potential consequence of fat malabsorption is diarrhea with poor absorption of fat-soluble medications and vitamins. The effects of long-term orlistat therapy in obese patients have not been thoroughly evaluated.

**Growth hormone**, although not approved or typically thought of as a drug in the treatment of obesity, does in indi-

viduals with PWS increase muscle mass and frequently increases allowable calories. However, insurance companies will only approve the use of growth hormone in PWS when there is short stature and demonstrable growth hormone deficiency. The costs to families paying for the growth hormone therapy themselves is prohibitive (\$30,000 to \$40,000 per year).

No current available therapy addresses the central mechanisms (i.e., controlled by the brain) regulating body weight. Various pharmaceutical companies and academic institutions are presently investigating different hormonal/neurotransmitter therapies using such agents as leptin analogs and NPY antagonists. Clinical research trials using leptin have just recently begun for the treatment of obesity in adults. However, it is doubtful whether giving leptin as a medication for the treatment of obesity in people with PWS would be helpful since the leptin levels are already high in these individuals.

### Surgical intervention

Various surgical treatments have been used in the obese population, including jejunoileal shunts and gastroplasty (reduction in the size of the stomach), but these have rarely been used in individuals with PWS and cannot be recommended in the PWS population due to the central nature of obesity in PWS.

### Summary of treatment strategies

Prevention of obesity through early diagnosis, education, and treatment is the best approach, particularly in the PWS population, but this is not always possible. Many of our patients already come to us morbidly obese and treatment plans need to be devised. This typically involves a combination of diet, exercise, and behavior management. Medication is typically added to the regimen if a significant weight reduction has not occurred after six to 12 months of diet and exercise, and if the BMI is greater than 30. However, there are presently no quick pharmacologic fixes for obesity and there may never be any. No treatment can be successful without a change in lifestyle (i.e., diet and exercise). A true weight loss program in the home setting is difficult, but it is achievable with the right guidance and a whole lot of dedication on everyone's part.

We have been very successful working with the Gainesville ARC group homes in achieving weight reductions in

all our 21 individuals with PWS for several reasons:

- 1) There is a very motivated and experienced staff who are knowledgeable in PWS and behavior management;
- 2) There is a strict adherence to the diet and a constant monitoring of the individual to assure compliance; and
- 3) There exists a dedicated exercise program.

Medications such as Prozac have been helpful, not so much as an appetite suppressant but in making the individual with PWS more malleable and compliant with the behavior management program. All 21 individuals in our program have lost a significant amount of weight, typically in the 50-100 pounds per year range. Those individuals that have been in the group homes for several years are all out of the obese range, with BMIs of 21-26.

## Concluding Thoughts

It is interesting to note in our patients (PWS and PWS-like) the high concordance of early obesity (starting at less than 3 years of age) and developmental delay. It may be that a defect in the regulation or the expression of particular neuropeptides leads not only to obesity, but also to learning impairment. It is well known that high levels of natural compounds in the body can lead to mental retardation (e.g., high levels of phenylalanine leading to PKU, high levels of galactose leading to Galactosemia, high levels of ammonium leading to Urea Cycle Defects). Therefore it is reasonable to speculate that high levels of various neurotransmitters or other hormones (e.g., leptin, neuropeptide Y, galanin) at a young age could lead to both obesity and learning impairment. This is understandable in the context of our understanding

that brain cells stop dividing at about 5 years of age and that nerve sheaths are fully myelinated by 5 to 8 years of age. Furthermore, there is anecdotal data from several groups and one published report that young children with Prader-Willi syndrome do better cognitively with early dietary control.

The last several years have shed a great deal of light on the cascade of events that lead to obesity and weight control. However there is a great deal still to learn. A combination of human and animal research studies will aid in our understanding and lead eventually to better treatments for obesity, not just for those with PWS, but for other conditions as well.

## BMI Reference Chart

To determine the approximate body mass index (BMI) of an adult with PWS, find the person's height in inches in the left-hand column of the chart below, go across that row to the figure that is closest to the person's weight in pounds, then go up to the number in the shaded box directly above that weight to find the BMI. A BMI of 21-26 is ideal; over 27 is considered obese.

For a more precise calculation, or for heights and weights not shown on the chart, compute BMI as follows:

- 1) Multiply weight in pounds by 0.454 to convert to kilograms.
- 2) Multiply height in inches by 0.0254 to convert to meters, then multiply the result by itself to get the square of the meters.
- 3) Divide the result of Step 1 (weight in kilograms) by the result of Step 2 (height in meters, squared).

Body Mass Index (BMI) for selected heights and weights																					
	20	21	22	23	24	25	26	27	28	29	30	31	32	33	34	35	36	37	38	39	40
Height (inches)	Weight (pounds)																				
	← Obesity Range →																				
56	89	94	98	102	107	111	116	120	125	129	134	138	143	147	152	156	160	165	169	174	178
57	92	97	102	106	111	115	120	125	129	134	139	143	148	152	157	162	166	171	175	180	185
58	95	100	105	110	114	119	124	129	133	138	143	148	152	157	162	167	172	176	181	186	191
59	99	104	109	114	119	124	129	134	139	144	149	154	159	164	169	174	179	184	188	193	198
60	102	107	112	117	122	127	132	138	143	148	153	158	163	168	173	178	183	188	194	199	204
61	106	111	117	122	127	132	138	143	148	154	159	164	169	175	180	185	191	196	201	207	212
62	109	114	120	125	130	136	141	147	152	158	163	168	174	179	185	190	196	201	206	212	217
63	113	119	124	130	135	141	147	152	158	164	169	175	181	186	192	198	203	209	214	220	226
64	117	123	129	135	141	146	152	158	164	170	176	182	187	193	199	205	211	217	223	228	234
65	120	126	132	138	144	150	156	162	168	174	180	186	192	198	204	210	216	222	228	234	240
66	124	131	137	143	149	156	162	168	174	180	187	193	199	205	212	218	224	230	236	243	249

# Reflections On Our First Two Years With Prader-Willi Syndrome

by Rachel W. Tugon, Louisville, Kentucky

**T**here was a time, not so long ago, when seeing “PW” meant seeing my father’s initials. It’s strange how something so familiar can take on new worlds of meaning in the blink of an eye. A little over a year ago, I heard the words Prader-Willi syndrome for the first time. Now I hear them all the time, when I’m awake, when I’m asleep and especially in that twilight zone where reality tends to blur. These words that first prompted the response, “Prader-What?” are now woven into the fabric of my family’s life and future.

Two years ago, God gave us a most special gift, our precious daughter, Erin. Having her sister, Kristen, five years earlier had hopelessly converted us to parenthood. We were thrilled to find that we were having another girl because our first had been so much more wonderful than we could have ever imagined. There were some complications in the pregnancy but we just kept saying that if we could just get through this, all our ducks would be in a row. Well, one of them must have escaped.

Erin was born on Feb. 8, 1996. Within two hours, we knew that something was wrong. Her muscle tone was so low that she looked like a rag doll when you picked her up. She slept constantly, couldn’t suck and had no response to pain. Instead of being placed behind the happy window where the families get to admire their newborn treasures, we were invited to the party in neonatal intensive care. While there, Erin was tested for everything imaginable, but nothing came back to give us any answers or direction. When she was able to take a little milk from a bottle, we were sent home with only a new diaper bag and the diagnosis of hypotonia, or low muscle tone.

After we got home, Erin continued to sleep. We had to wake her to feed her. It took two people to provide enough stimulation to keep her awake long enough for her to drink two ounces of milk. Because she was so quiet, people

would always remark about what a “good” baby she was. Although there was never any question about her goodness, in my heart I knew that whatever was going on with Erin had nothing to do with her being good.

While she was hospitalized at two months with pneumonia, she saw a geneticist. He ordered tests for something of which I had never heard, Prader-Willi syndrome. A nurse gave me a fact sheet on this and it sounded so terrible that I had to put it away. At the time, we had all we could handle with the pneumonia and it was just too much to think about. After three weeks, the test came back negative, and we were so relieved not to have PWS as a possibility. We still didn’t know what she had, but we thought we knew what she didn’t have.

At six months, the pediatrician recommended starting Erin on solid food, so we began with cereal and typical baby food. Up until this time, Erin had been small and weighed only 12 pounds at six months. Within three months after she started eating, she gained 10 pounds, almost doubling in size. The bizarre thing was that she didn’t eat that much. She never finished all that the pediatrician recommended she eat, but she was rapidly gaining weight. Her doctor said that it was just a growth spurt and not to worry, but by the third month we were more than worried. I cut back

on the food we offered her, and even on the lesser amounts she still gained weight.

It was now time for our follow-up visit with the geneticist. After we told him the story of her weight gain, he recommended that she be retested for Prader-Willi syndrome. I could tell by the look on his face that there was reason for concern. Deep inside, I already knew what the test would say.

All too quickly, the tests came back and our worst fears were confirmed. Erin had tested positive for the more rare form of PWS. Usually, PWS comes from a deletion on chromosome 15. Her initial test had come back with a complete chromosome and thus a negative test. The second test, newer and more conclusive, showed that Erin’s PWS came from uniparental disomy. This is where both



Erin Marlow Tugon, age 2, frolicking with her preschool buddies.

chromosome 15s are from the mother instead of one from each parent as would normally be the case.

Although we had some warning, the diagnosis was devastating. The materials the doctors presented to us painted a gruesome, dismal future. In those days, it felt like we were wandering around in a thick, dark fog. This is the kind of thing that happens to other people's children, not ours. Family and friends tried to be supportive, but their dependence on miracles and misdiagnosis felt so hollow. Every time I looked into Erin's beautiful blue eyes, I felt my heart break again and again. Parents are supposed to fix things for their children. I'd never felt so helpless as then, feeling that there was something so important in her life that I couldn't fix.

**A**fter the shock of the diagnosis began to ease, we started to research this syndrome and break it down into its separate parts. She had been in physical, occupational, and speech therapy since she was three months old. We added a dietitian, cranio-sacral therapy, a family counselor to give us guidance on behavioral management, and developmental intervention to her cadre of doctors and specialists. Joining our local support group, Prader-Willi Syndrome Association of Kentucky, helped us feel less alone. Although no one can truly know another's pain, we did know that we were among people who had had similar experiences and could understand and empathize with our confusion.

The dietitian helped us come up with an eating plan that would allow Erin the nutrition necessary for growth while at the same time restricting her calories. After some experimenting, we found that Erin could handle 600 calories a day and not gain weight. That was just about 300 calories plus her milk. Prader-Willi brings a very slow metabolism, an insatiable appetite, and low muscle tone. That combination makes it very tricky to plan your child's eating. I measure every bite of food she has and I even count her Cheerios! As wild as that seems, Erin has gone from being way off the chart to the 25th percentile in weight.

At 20 months, she began taking growth hormone, as testing had shown her pituitary not to be producing sufficient growth hormone (GH). We have seen dramatic effects since beginning this therapy. In addition to an increase in length, Erin has lost weight, shown a better distribution of weight, has increased metabolism which has allowed

**“ ... we have served notice to PWS that it is in for the fight of its life. We may not be able to make it go away, but we can take each of its symptoms, one by one, and set up a plan of attack.”**

an increase in calories, and has increased her muscle mass. All this has led to an increase in strength and endurance which makes it easier for her to get more exercise. People who have not seen her since she began growth hormone therapy have said that they would hardly have recognized her as her face as well as her body has changed. We had questions about GH at first, but so far we are very pleased with the results.

We are so very thankful for an early diagnosis. By trying to develop good eating habits now, we hope to ease the pain of Prader-Willi syndrome for her later. We can control her eating now but that will not always be the case. Our hope is that because she has always eaten this way, it will seem normal to her. I have fought a losing (no pun intended) battle with weight all of my life and I know how hard it is to change patterns. If it's this hard for an adult who understands the consequences, how much more for a child who just knows that she is so very hungry and just can't get full, no matter how much she eats.

**M**y husband and I have developed a defiant attitude about

PWS in the last year. We see PWS as an uninvited, unwelcome guest in our house. This gives us something outside Erin to channel our negative feelings toward. In a sense, we have served notice to PWS that it is in for the fight of its life. We may not be able to make it go away, but we can take each of its symptoms, one by one, and set up a plan of attack. We get almost corny at times with our “If it's a fight you want, it's a fight you'll get!” attitude, but it's helping us find our way through this. Right now, Erin is doing beautifully. She began to walk at 21 months and is talking up a storm. It seems there is no stopping her now, not that we'd want to! You'd have to see her to understand this, but she is a stunningly beautiful child. Although I will admit to being shamelessly proud, people are constantly telling us what an absolute doll she is. She has worked so hard and come so far. We will do whatever it takes to give her the best chance possible.

**N**ow that we have had some time to adjust to the diagnosis and to learn more about the syndrome, we are getting a better perspective on our life and future. Yes, Erin has a rare, genetic birth defect. Yes, she will face many challenges and difficulties. Yes, PWS has changed all of our lives forever. And we couldn't be happier! Don't get me wrong ... I'd give or do anything to make this thing go away for her sake. But for us, we are just so thankful that God has given us this precious little girl who has brought nothing but love and laughter into our lives. She has opened our eyes to a whole new world, and through her we have learned so much. I have caused my share of trouble during my life, but somewhere, sometime, I must have done something right because God allowed us to have these two wonderful daughters, both with unique talents and challenges. We are so thankful that they both chose us to be their parents and are doing our best to be worthy of them. Prader-Willi or no Prader-Willi, our job as parents is to help our children be as healthy and happy as possible. Nothing more and nothing less.

### Too Subtle Symptoms

Among the many concerns we face as parents of people with PWS, closely monitoring the day-to-day health of our kids has proven to be a surprisingly challenging task. Ordinary childhood illnesses, easily detected in non-PWS kids by fever, vomiting, or pain are often masked in our children by their high tolerance of pain, lack of a vomiting instinct, or lowered body temperatures.

I have learned to look for the little things in my 5-year-old daughter's behavior: Mildly labored breathing and a series of unfinished snacks resulted in a diagnosis of strep throat. Increased lethargy was an ear infection. Never a fever. No vomiting or complaints of pain. Just a mother's awareness of the subtle changes in her child.

Yes, I have learned discernment. But how to teach it to others? That is the challenge (frustration!?) before me as my precious little child grows. To teach her doctors that the lack of a fever or pain doesn't always mean a lack of illness. Or to make her relatives, teachers, and other caregivers understand that food left on the plate, perfectly normal behavior in other children, might be indicative of something more serious in my child.

—Parent, Springfield, Virginia



*Editor's note:* This serious issue of the lack of pain signals in people with PWS was addressed by several presenters at the recent international PWS conference:

A poster presentation by Dr. Robert H. Wharton of Harvard Medical School in Boston gave the first confirmation, based on parent surveys, of an atypical response to pain and stress in Prader-Willi syndrome. An analysis of the reports from 16 families on their children with PWS found that in infancy 14 had no colic or response to colic and 13 had no response to teething; 9 of the 16 had no response to blood tests.

A report by Astrid Schulze and others (Denmark) on a young adult's death from severe illness preceded by no signaling pain "highlights the need for parents, carers, and medical staff to be aware of subtle changes in the behavior patterns of people with PWS. Such change(s) in behavior may be indicative of the early stages of disease onset at a time when treatment intervention may be effective."

PWSA's "Medical Alert" brochure is a tool parents can use to alert medical professionals about this problem. The brochure can be downloaded from the PWSA Web page or ordered from the national PWSA office.

### Dear Janalee,

Thanks SOOOOO much for posting the policy statement on "Adults With Prader-Willi Syndrome and Decisions Regarding Least Restrictive Environment and the Right to Eat" [on PWSA's Web page, <http://www.pwsausa.org>]. Now that this message is backed by the authoritative voice of PWSA (USA), perhaps we local advocates will be taken seriously, and not seen as some diet-obsessive fanatics who want skinny kids.

I recently pulled my daughter, Karie, out of her day program because they were not willing to comply with their prior commitment to provide close supervision, making arguments for "personal rights" and freedom of choice. Karie had been slowly gaining weight from food acquired at this program, and when she went AWOL one day and the staff LET HER GO, I pulled her from the program. (She was gone for four hours, wandering downtown, until a special police dispatch team found her.) Now she is in a program with strict food control and close supervision, and has lost almost 10 pounds in the past two months, thanks to a caring, conscientious, and cooperative team.

We appreciate all the information that is available at your Web site, and the personal one-on-one support you have provided to parents. Continued communication and education will raise awareness and result in an improved quality of life for our special children.

Teresa Kellerman  
President, PWSA of Arizona

### Parents! We Need To Hear from You

The national PWSA office gets calls on several topics for which we have little printed information. We would like to collect parent/caregiver wisdom and experience on these topics to share with our members and callers. Please let us hear from you by mail (see p. 2 for national office address), fax (941-312-0142), or e-mail ([PWSAUSA@aol.com](mailto:PWSAUSA@aol.com)).

- ◆ **Locks and alarms** — What are the most effective ways to lock up food? We'd like specific information on types of locks and alarms, including brand names and photos or diagrams if possible, and their installation on various food sources, e.g., refrigerators, cabinets, pantries, kitchen doors. What tips do you have for effective use of locks?
- ◆ **Toilet training** — Toilet training is commonly delayed in toddlers who have PWS or any other developmental disorder. Tell us in detail about your experience in toilet training your child. What worked and what didn't? What about night-time training? When and how was that accomplished? What references or products were helpful for you? How have you handled toileting issues in the daycare or school setting?
- ◆ **Nutrition for infants and toddlers with PWS** — PWSA is planning a booklet that will offer practical advice on early feeding, stressing the importance of providing adequate nourishment for the child while avoiding obesity. In order to cover the topic to meet parent needs, we need to hear your personal concerns and helpful suggestions. Carolyn Loker, the mother of a 3-year-old with PWS, will be developing this booklet, with the help of medical and nutrition experts. Please send submissions on this topic directly to: Carolyn Loker, 2213 Cross Country Dr., Kalamazoo, MI 49009, or e-mail her at [carolynloker@yahoo.com](mailto:carolynloker@yahoo.com).

# Twin Joys— and Movie Stars, to Boot

by Lisa Stover, Severna Park, MD

Yes, I'm a mother of twins with Prader-willi syndrome, but don't say "you poor thing" yet. In fact, don't say "you poor thing" at all, because that phrase simply has no place or truth in my life or the lives of my identical twin sons. Hunter and Scout were a normal twin pregnancy, I guess. I had no problems with them, no bed rest, and in fact worked up until the week before I delivered them. There were no real problems at birth or directly after, except for the feeding issue of them not wanting to eat, and this definitely was a problem. My husband and I would sit and try to force even just an ounce of milk into our sons, and it was very frustrating and very upsetting. They were being weighed almost daily at the doctor's office, and no one knew what the problem might be.

After a few months of this, we finally hospitalized and tube-fed them just to give them a "jump start." A few days ended up being two weeks, and every test was done on them, even a full genetic work-up. At discharge the only thing we had gotten out of our stay was a few pounds, which was wonderful, and a diagnosis of neonatal hypotonia (low muscle tone), failure to thrive, and urinary tract infections. But, no diagnosis of what the real problem might be or if there was one. After a trip to the local medical library a few weeks later to look up neonatal hypotonia, my husband and I asked to be referred to a doctor for testing for Prader-Willi syndrome. Of course, we had the testing done, and sure enough they both tested positive. We were relieved to know what was wrong with our children and disappointed that we, not the endless array of doctors we had dealt with, managed to diagnose our own children.

I won't kid you, the first few months were very hard, for me especially. I guess I went through what every parent does—grief, anger, sadness. My sons were only about 8 months old at the time they were diagnosed with the syndrome. The next six months or so was a time of learning, reading, and quite frankly being scared to death of what the future would hold for my children. Of course, they are only 2½ now, but as someone told me way back then, don't always take what you read as a "must be" for you. I know now that what that person told me was the truth. I know I have a long way to go with my kids and



Hunter and Scout Stover, now 2½, shared the role of the Amish infant in Tim Allen's recent movie, "For Richer or Poorer," released last December. Below: Tim Allen and young co-star in costume.

yes, there will probably be some things to deal with that I would rather not, but we're just taking it one day at a time. My sons are such a blessing to me and my husband. We thank God each day for them, just as they are. They are such a delight to us with their personalities, their smiles, their laughter and their desire to accomplish things that come easy for "normal kids."

And you may think that having two with the syndrome is difficult, because you know how you feel with just one child, but like I always tell people, I thank God that both my sons have the syndrome. They won't be labeled as the "normal" child and the "challenged" child, especially when they are identical; one won't feel bad because the other can do things that he can't do; and especially because they will always have each other to comfort, lean on, and to confide in. I can say I sympathize with how they feel or what they're going through, but they will truly have someone who knows, and not just some other person with the syndrome, but someone they've grown up with, their brother and best friend. I can't tell you how thankful I am for that.

Hunter and Scout are not only good little boys, they are also so beautiful, like little angel faces. We get stopped everywhere we go by people who can't get over how beautiful they are and who tell us they should be in movies. Well, they *are*, and if you don't believe me just go to your local video store and rent "For Richer or Poorer" (1997) with Tim Allen and Kirstie Alley. My sons, my sons with Prader-Willi syndrome, play the only baby in the movie



and are in about four scenes. They play the baby of the Amish family that Tim and Kirstie hide out with.

Out of 27 sets of twins who auditioned for the part, my sons got it. Everyone couldn't get over how beautiful, sweet, and special they are. They were the most well-behaved kids that they had seen, and that's one of the reasons that they got the part. They filmed for six weeks and we all had a great time. Everyone was really great to them, and Tim and Kirstie played with them and both said how wonderful it was to work with them. What a great experience!

All I want is to do the best job I can and make my sons the best people they can be, to give them every opportunity to succeed and be productive. Isn't that what any parent wants? Yes, it takes a lot. A lot of time, patience, love, and hoping for the best and striving for that, not dwelling on the worst. But, really, what more important thing do I have to do? Nothing.

*Editor's note:* Lisa can be reached by e-mail at: Pwsmom2@aol.com

## The Leeza Show

On May 26th, the nationally syndicated daytime talk show host, Leeza Gibbons, did a one-hour program on Prader-Willi syndrome. It featured two families who have daughters with PWS (see photo, below), PWSA Executive Director Janalee Heinemann, Board Member Ken Smith (program director at The Children's Institute in Pittsburgh), and Marlene Corrigan and her attorney, Michael Cardoza. Marlene had been charged with a felony in the death of her 13-year-old daughter, Christina, who weighed 680 pounds when she died and may have had PWS. The case has received international coverage and has been responsible for significant interest in PWS in this country. Several other PWSA families also were in the audience and were interviewed, including Board Member Fran Moss and her daughter Melissa. Leeza was very sensitive in her interviewing and covered several issues related to PWS besides the insatiable appetite — behavior problems, skin picking, the emotional pain of being ridiculed, etc.



Melanie Billigen and Kristi Pickert share an informal moment with Leeza behind the scenes.

PWSA (USA) received over 500 calls in the first three days after the Leeza show aired! Numerous children have been diagnosed thanks to this show and to the vast amount of other publicity we have received as a spin-off of PWSA's involvement in the Corrigan court case, much of which was detailed in our March-April newsletter. Katie Walker and her parents, from Oklahoma, who were featured in the Extra show back in February, had another TV appearance in early June on Sally Jessie Raphael's show, and there have been more articles in major newspapers. Awareness of the syndrome is at an all time high. It touches our hearts every time a parent calls and says "I have felt so all alone," and is moved to tears when they realize they are not to blame for their child's problems.

Besides the heightened awareness this show brought regarding the syndrome, we have received approval from Paramount Pictures to distribute videotapes of the Leeza Show for educational purposes. Since it describes some rather dramatic behavior problems, it would not be appropriate for those dealing with young children with PWS, but it could be very effective for parents who need to verify for schools, etc., that the weight and behavior problems are caused by the syndrome — not poor parenting.

—Janalee

(See ordering information for the Leeza videotape on page 15.)

## Crisis, Grief Programs Under Development

*Thanks to Saint Boniface Episcopal Church* in Siesta Key, Florida, PWSA (USA) was awarded a \$2,000 grant to assist in the reproduction of our dramatic "refrigerator" folders for new "*Crisis Intervention Packets for Families in Distress.*"

If you are a member and dealing with a legal, SSI, or placement crisis, we can now provide a powerful support packet. Call the national office for details.



*"Supporting the PWS Bereaved Parent"* is a one-year bereavement follow-up program conceived by Executive Director Janalee Heinemann and presented at the IPWSO conference in Italy. It is designed to support families who lose a child with PWS by sending them a series of five mailings during the year following their loss, and connecting them with volunteers on a special sharing list if they wish to talk with someone about their experience.

Janalee would like to pursue a grant to implement this program for all of our PWSA families who have lost a child. Her goal is to have volunteer parents involved with the implementation. If anyone is aware of potential grant funding or is interested in volunteering for this program, please contact Janalee.



*A Special Thanks to the George Edward Durell Foundation* of Winchester, Virginia, for awarding PWSA (USA) an unrestricted grant of \$1,400.

## Text of PWSA's new wallet-size card for restaurant staff:

\* ATTENTION WAIT STAFF \*

An individual you are serving has Prader-Willi syndrome (PWS). Children born with this syndrome have an insatiable appetite due to a wrong message to the brain that always triggers "hunger," never "full." They also gain weight with fewer calories. Because of this, these individuals are kept on a very strict diet to avoid life-threatening obesity. We respectfully ask that you not offer extras such as bread, butter, desserts, or even extra drinks, unless they are diet. We appreciate your cooperation.

## Many New Products Available from PWSA

PWSA national office staff, board members, and committees of the board have worked furiously over the past year to bring members a number of new and useful information products. PWSA's brand new video, professionally created to introduce all types of audiences to Prader-Willi syndrome, made its debut at the PWSA national conference in Ohio and is now available for sale. In addition, just before the conference, PWSA received permission from Paramount Pictures to distribute copies of the Leeza Gibbons show on PWS, which was perhaps the most balanced and informative talk show yet produced on the syndrome.

The PWSA publications committee has produced several new—and much-needed—compilations of articles in booklet form, covering exercise, behavior, and planning for the future; and an arrangement with the publisher of *Prader-Willi Perspectives* allows us to offer four titles listed at right for sale through PWSA.

Two practical tools have just been developed, in the form of wallet-sized cards, to help families quickly educate restaurant staff when dining out and law enforcement authorities in the event of a crisis.



**This page (or a copy of it) can be used to order these new products.**

Please write your name and address below and send payment in the form of check, money order, or MasterCard/VISA information. (See box for shipping charges.)

Name \_\_\_\_\_

Address \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

Order Amount \$ \_\_\_\_\_

Shipping \$ \_\_\_\_\_

Total \$ \_\_\_\_\_

Charge:  MasterCard  VISA

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### Add Shipping Charges:

\$1.50 for orders up to \$5.99  
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 \$10.50 for orders of \$100.00 & up



## PWSA Products

### Videos

— **New PWSA (USA) Video: "Searching for Solutions"**

A 16-minute introduction to PWS, created by award-winning producers and graphic artists. **\$20 members/\$25 nonmembers**

— **Leeza Show on PWS, broadcast 5/26/98: "Food as an Ultimate Passion and Deadly Curse."**

Interviews with families and PWSA officials cover a broad range of issues—weight, behavior, high pain threshold, etc. (Not recommended for parents of young children.) Distribution authorized by Paramount Pictures Corp. Suggested donation to defray costs of copying videotapes: **\$20 members/\$30 nonmembers**

### Booklets

— **Estate Planning, Wills and Trusts** — A collection of articles and resources by William L.E. Dussault and others. **\$6.00 members/\$8.50 nonmembers**

— **Behavior Management** — A collection of articles from *The Gathered View* and other sources. **\$6.00 members/\$8.50 nonmembers**

— **Exercise, Activities, and Crafts**

Articles from *The Gathered View* and other sources, with recommendations from Jennifer Deau, exercise specialist at The Children's Institute in Pittsburgh. **\$6.00 members/\$8.50 nonmembers**

— **Growth Hormone** — A collection of articles. **\$3.00 members/\$5.50 nonmembers**

### Prader-Willi Perspectives Information Series

— **The Child With Prader-Willi Syndrome: Birth to Three**

Booklet by Robert H. Wharton, M.D., Kaen Levine, Ph.D., & Deirdre C. Mulcahy, M.S. **\$5.00**

— **Children with Prader-Willi Syndrome: Information for School Staff** (Third Edition, 1996)

Booklet by Karen Levine, Ph.D., Robert H. Wharton, M.D. & Margaret E. Comeau on learning and behavior issues and educational testing. **\$5.00**

— **Physical Therapy Intervention for Individuals with Prader-Willi Syndrome** — Booklet by Maria T. Fragala. **\$4.00**

— **Transition From School to Adult Services in Prader-Willi Syndrome: What Parents Need To Know** — Booklet by Julie A. Seguin, M.S., CRC & Robert M. Hodapp, Ph.D. **\$10.00**

### Wallet-Size Information Cards

— **Restaurant cards**

Designed to be handed to staff waiting on tables, this card gives a brief description of PWS and requests that no "extras" be offered at your table. **\$2.00 for pack of 20**

— **Law enforcement cards**

Designed to be given to law enforcement personnel in the event of a behavior crisis involving someone with PWS, this card explains characteristic behaviors and requests support and understanding. **\$1.00 for pack of 4**

Mail this form along with your name and address and a check or money order, including correct shipping charges (left), to: **PWSA (USA), 5700 Midnight Pass Rd., Suite 6, Sarasota, FL 34242**

**“Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it’s the only thing that ever does.” — Margaret Mead**

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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.

