



# The *Gathered View*

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

September-October 2001  
Volume 26, Number 5

## 2001 World Conference

# We Join Hands Together And Love Makes Us Fly

## *"Fly High"*

Lyrics by Linda Thornton  
Music by Giorgio Fornasier  
Arranged by Roberto Antonello

We come here together, to meet with each other,  
To share our life stories, to tell of our glories;  
Sometimes it's not easy to always be different,  
But you are my friend here and you understand me

We're brothers, we're sisters, we're part of the family  
That binds us together from coastland to city;  
Like droplets of rainfall, we flow like a river,  
Combining our wisdom like powerful sea

So let us stand tall now, let's stand tall with pride.  
Let's celebrate life now, both your life and mine;  
We stand on the same side, 'though we live apart,  
Just knowing you're there, friend, stays deep in my heart

REFRAIN (repeat after each verse)

Across the world's oceans, across the world's skies  
We join hands together and love makes us fly — high!



*Giorgio Fornasier of Italy and Linda Thornton of New Zealand performing 'Fly High' for the first time at the 2001 World Conference.*

Dear Friends,

"Fly High" is not my song, but our song, purposely written with four hands by two parents of children with PWS.

We can be proud of how the Conference went. We keep receiving compliments and enthusiastic messages from every corner of the world. You worked very hard, but I am sure you can be very satisfied of the success of the event you built up.

Ciao,  
Giorgio

*Conference continued on pages 8 & 9*

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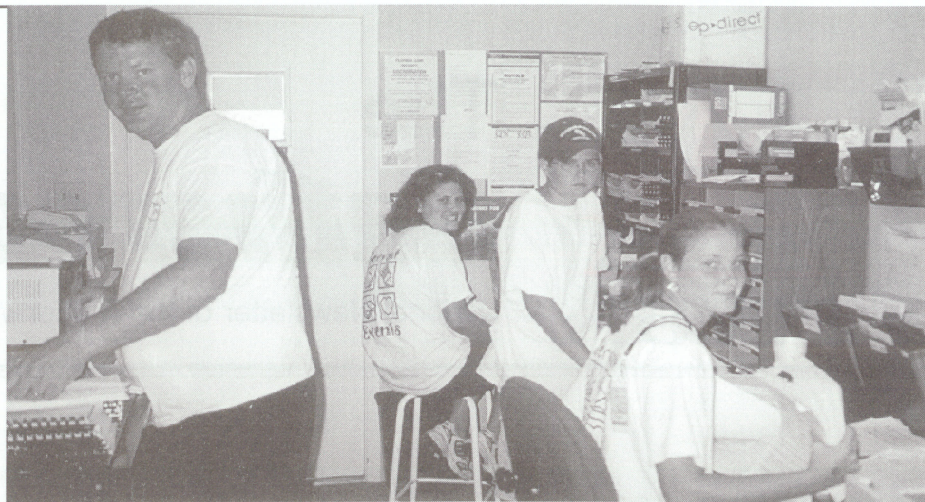
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*The Gathered View* welcomes articles, letters, personal stories and photographs and news of interest to those concerned with Prader-Willi syndrome.

Communications regarding *The Gathered View* or PWSA membership and services should be directed to the national office of PWSA (USA) in Sarasota, Florida at the address above.



### Thanks a million

*Our thanks to the 20 young volunteers from the Concordia Lutheran Church Mission Group of Sarasota who came to help out at the PWSA (USA) office in July.*

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# Sen. Dole PSA for PWS to Air This Fall

Sen. Bob Dole filmed a public service announcement (PSA) about Prader-Willi syndrome on August 2, 2001 in Washington, D.C. at the Republican Party Studio. Appearing with him is Amanda Diaz, age 7, who has PWS.

Susan Wheeler, communications director for U.S. Senator Crapo of Idaho, coordinated the filming. Susan is Pamela Tobler's sister. Pamela is on the PWSA (USA) national board of directors, serves as public relations liaison and is the mother of Nathan, who has PWS.

The PSA is expected to air this fall and will also appear on the PWSA (USA) web site, [www.pwsausa.org](http://www.pwsausa.org).

Thanks to funding from Pharmacia, tapes will be made in the video format needed for each TV station. Nuvidia in Overland Park, Kansas is donating all the tapes needed.

Satellite link time will be purchased for those TV stations willing to up link to the satellite and dub their own tapes.



*Sen. Bob Dole and Amanda Diaz*

After TV stations receive the tape, it will take 6-8 weeks before the public service announcement is put into their rotation.

## Prader-Willi Clinic is Opening in Georgia

PWSA/GA is pleased to announce that the Prader-Willi Syndrome Program of Georgia will hold its first clinic day on September 19 at Hughes Spalding Children's Hospital in Atlanta.

The PWS Program is a collaboration of medical and educational professionals committed to advancing the knowledge, understanding and treatment of people affected by PWS. The first step is to establish a PWS clinic in Georgia.

The clinic will be held one full day five times a year for children up to age 18 on Sept. 19 and November 7 in 2001, and January 16, March 6 and May 29 in 2002.

Adults will receive care at a separate clinic that is scheduled to open next year at a different location.

Dr. Randell Alexander is medical director of the clinic. Dr. Alexander is co-editor of the textbook, *Management of Prader-Willi Syndrome*, and helped establish one of the first PWS clinics in the United States at the University of Iowa. He now lives in Atlanta and teaches at Morehouse School of Medicine.

Associate Medical Director Dr. Leslie Rubin has also played a crucial role in developing this program. Dr. Rubin is director of the Division of Developmental Pediatrics and associate professor of pediatrics at Emory University Hospital in Atlanta. He also works with the CP clinic at Hughes Spalding, where the Prader-Willi clinic will be housed.

To assist in the direct assessment, evaluation and medical care that they will be providing each patient, Drs. Alexander and Rubin have enlisted specialists for the clinic in the following areas: nutrition, behavior/ education, physical therapy, occupational therapy and speech/language therapy.

Patients will be able to access dental, genetic and endocrine specialists through the clinic as well. Administrative, nursing and family support services will also be available.

Medicaid and private insurance will be accepted and handled by Hughes Spalding.

The Georgia PWS Association will contribute to the operating expenses and is seeking donations to support some of the clinic specialists.

"We are very excited that this 15-year-old dream is becoming a reality," said Hope Mays, president of PWSA/Ga. "We expect great things to come about as a result of this program: Increased

awareness and diagnoses, better understanding of the syndrome by professionals, communities and families and improved services for all individuals with Prader-Willi Syndrome."

## Board Members Elected

Three incumbents and one newcomer were elected to the PWSA (USA) Board of Directors in June, 2001. They each will serve 3-year terms.

New to the board is Robert Lutz of Cincinnati, Ohio. He and his wife Debra are the parents of Isabel, who has PWS. Currently the chief financial officer of Cinergy Corp., Rob brings his business and management expertise to assist PWSA (USA).

Daniel J. Driscoll, M.D., Ph.D. of Gainesville, Florida, is a professor of pediatrics and molecular genetics at the University of Florida College of Medicine. Dr. Driscoll is co-chair of PWSA (USA)'s Clinical Advisory Board and co-chaired the Scientific Conferences at the 2001 PWSA/IPWSO World Conference.

James P. Gardner of White Bear Lake, Minnesota is proprietor of Gardner Management Co. He has served on the Board of Directors since 1995. He and his wife Joan co-chaired the 2001 PWSA/IPWSO World Conference. Jim and Joan are the parents of Lawrence, who has PWS.

Ken Smith of Pittsburgh, Pennsylvania is program manager at the Children's Institute in Pittsburgh, and has worked with people with PWS and other disabilities for 16 years. Ken, who is now board chair, will be serving his third term on the board.



# PWS and Obesity, and PWS look-alikes

By *Celanie K. Christensen, M.S. and Bryan E. Hainline, M.D., Ph.D.*

*In the past, obesity has not been considered a true medical problem, but a sign of psychological weakness. Recent research has led to a paradigm shift in the medical management of obesity. Prader-Willi syndrome has become a more commonly considered genetic syndrome in the evaluation of an obese child.*

*In this article, Celanie Christensen and Dr. Bryan E. Hainline discuss PWS and other genetic syndromes that share similar characteristics.*

*Celanie K. Christensen graduated with distinction in 1999 from the University of Iowa, double majoring in Biologi-*

*cal issues associated with obesity. Our society stigmatizes obese individuals, and poor self-esteem is a common finding. When a child is evaluated for obesity, the list of possible diagnoses that the doctor considers should include genetic syndromes, especially when unusual physical features, mental retardation, or other health problems are present.*

*Obesity is a multifactorial (interaction of many causes) condition with both genetic and environmental components.*

*One way to quantify obesity is by calculating an individual's Body Mass Index (BMI). BMI can be calculated by dividing weight in kg. by height squared*

*sumption and energy expenditure.*

*However, rare exceptions to the accepted multifactorial mode of inheritance do exist in monogenic (single gene) syndromes with obesity as a main phenotypic component.*

*There are many other possibilities that need to be considered in an obesity evaluation. Some are common genetic syndromes that are easily tested for, while others are more obscure.*

*Genetic syndromes with obesity and delayed/altered intellectual development as a major part of the phenotype include Prader-Willi syndrome, Albright hereditary osteodystrophy, Alstrom syndrome,*

**Most cases of obesity can be explained by a multifactorial mode of inheritance involving genes, food consumption and energy expenditure.**

*cal Sciences and Psychology. She received her M.S. in Medical and Molecular Genetics from Indiana University in 2001 and plans to pursue a career in genetic counseling.*

*Bryan E. Hainline, MD, Ph.D is Clinical Associate Professor of Pediatrics and Assistant Professor of Medical and Molecular Genetics at Indiana University School of Medicine. He is affiliated with the J.W. Riley Hospital for Children in Indianapolis, Indiana. Dr. Hainline is also a member of the PWSA (USA) Clinical Advisory Board. - Ed.*

Obesity is a common finding in our general population, the proportion of obese individuals having increased at an alarming rate all over the world in the past 20 years. Compared to growth failure, obesity is rare in a population of individuals with a genetic syndrome. One database for genetic syndromes, Online Mendelian Inheritance in Man, contains 169 matches for obesity and 421 matches for growth failure.

Obesity is a medical problem that increases the risk for heart disease, diabetes (type II), high blood pressure, stroke, orthopedic problems, sleep apnea, and mortality from cancer. Besides medical issues, there are also psychologi-

cal issues associated with obesity. Our society stigmatizes obese individuals, and poor self-esteem is a common finding. When a child is evaluated for obesity, the list of possible diagnoses that the doctor considers should include genetic syndromes, especially when unusual physical features, mental retardation, or other health problems are present.

Obesity is a multifactorial (interaction of many causes) condition with both genetic and environmental components.

One way to quantify obesity is by calculating an individual's Body Mass Index (BMI). BMI can be calculated by dividing weight in kg. by height squared

in (m<sup>2</sup>) or if converting in the English system, BMI = [Weight in pounds ÷ Height in inches ÷ Height in inches] x 703.

Recent studies have shown that 54 percent of the adult population in the U.S. is overweight and 22 percent are obese. The youth of this country are also showing an increase in BMI: 25 percent of U.S. children can be classified as overweight or obese.

While there is a definite genetic component to obesity, a large part of the current epidemic is due to environmental factors that promote overeating and reduced physical activity. Humans have developed mechanisms to protect against weight loss in times of scarcity, but protection against weight gain in times of abundance is not as prevalent in the population.

Most cases of obesity can be explained by a multifactorial mode of inheritance involving genes, food con-

sumption and energy expenditure. However, rare exceptions to the accepted multifactorial mode of inheritance do exist in monogenic (single gene) syndromes with obesity as a main phenotypic component.

There are many other possibilities that need to be considered in an obesity evaluation. Some are common genetic syndromes that are easily tested for, while others are more obscure.

Genetic syndromes with obesity and delayed/altered intellectual development as a major part of the phenotype include Prader-Willi syndrome, Albright hereditary osteodystrophy, Alstrom syndrome,

Bardet-Biedl syndrome, Borjeson-Forssman-Lehmann syndrome, Cohen syndrome and fragile X syndrome. Chromosomal conditions, including Down, Klinefelter and Turner syndromes, also have an increased prevalence of obesity.

In recent years, several single gene mutations have been linked to extreme obesity. Certain forms of brain damage can also be a cause of extreme obesity.

Albright hereditary osteodystrophy (AHO) is characterized by variable mental retardation, short stature, round face, short neck, short bones of the fingers and toes, calcifications or ossifications in subcutaneous tissues often near joints, delayed development of the adult teeth, enamel hypoplasia and obesity.

Individuals with AHO can have obesity of prenatal onset or develop it within the first year of life. AHO is also known as pseudohypoparathyroidism type 1a. AHO can be diagnosed by testing the amount of calcium and phosphorus in the blood and the level of cyclic AMP in urine.

The gene for AHO has been mapped to chromosome 20, and it is inherited as a dominant condition.

*Hainline continued on page 5*



**Alstrom syndrome (AS)** produces symptoms in children that include progressive loss of vision and hearing, an unusual rash (acanthosis nigricans) that is seen in people with some types of diabetes, short stature, renal (kidney) failure, dilated or enlarged heart and obesity.

Visual impairment typically begins with photophobia (abnormal sensitivity to light) in the first 6 months and advances to blindness by 30 years. Obesity is typically present before 5 years and increases with age. Deafness usually presents late in the first decade, and renal failure often occurs by the third decade. AS is inherited in an autosomal recessive pattern, and one gene locus has been found on chromosome 2. Testing for AS is available on a research basis.

**Bardet-Biedl syndrome (BBS)** is characterized by more toes or fingers than normal at birth, progressive vision loss, renal (kidney) abnormalities, developmental delay, hypogonadism (lack of sexual development), short to low normal stature and obesity. Other findings include diabetes mellitus, endocrinological dysfunction, and behavioral abnormalities.

In BBS, the onset of obesity is usually in early childhood. Visual impairment usually presents with color and/or night blindness which progresses to blindness by 20 years. Renal abnormalities are varied, with 60 percent of patients having hypertension with renal failure occurring infrequently. BBS is inherited in an autosomal recessive pattern, and at least 6 gene loci have been found in different families.

Other families have not been linked to any of these loci, leading researchers to continue to look for other loci responsible for BBS. Currently, genetic testing is available on a research basis to families with one or more affected children.

**Borjeson-Forssman-Lehmann syndrome (BFLS)** is characterized by epilepsy, severe mental retardation, hypotonia, poor sexual development, swelling of subcutaneous tissue in the face, narrow palpebral fissures (narrow opening between the upper and lower eyelids), large ears, height below the 50th percentile, and obesity. BFLS is inherited in an X-linked recessive pattern.

An X chromosome gene, FGF13, has



*Bryan Hainline, M.D., Ph.D.*

**Many previous studies have used children seeking psychological or psychiatric treatment as a study population to observe differences between obese and non-obese children. It is very important that a correct diagnosis be made because of the varied treatment options available that are syndrome specific.**

been hypothesized as the gene responsible for BFLS. Males with BFLS are severely affected, and female carriers can be mildly to severely affected.

**Cohen syndrome** is characterized by mental retardation with a sociable/cheerful personality, small head size, hypotonia, abnormal curvature or rounding of the spine, short stature, narrow hands, progressive heart disease, near-sightedness, vision problems, prominent incisors, blood white cell problems, and obesity.

In Cohen syndrome, obesity tends to develop by the school-age years, and the mental handicap is non-progressive. Cohen syndrome is inherited in an autosomal recessive pattern. The Cohen syndrome gene is designated COH1 and is located on chromosome 8.

**Fragile X syndrome** is characterized by mental retardation, increased growth in childhood, large head, large ears, dental crowding, and large testicles. In some cases, obesity is also seen in individuals with fragile X syndrome.

Fragile X syndrome is an X-linked recessive condition involving the expansion of a repeated sequence DNA in the FMR1 gene on the X chromosome. There are three levels of repeats: normal, premutation, and full mutation.

Individuals with a premutation, especially females, have an increased risk of passing on a full mutation to offspring because of instability in the FMR1 gene.

A subset of patients with fragile X syndrome have a phenotype that is different from the classic presentation. These individuals have a full, round face, small and broad hands/feet, areas of more darkly colored skin (hyperpigmentation), and obesity. The reported phenotype in these patients has been compared to the PWS phenotype, with differences being lack of infantile floppiness and food seeking in childhood. In one study, individuals with these characteristics were found to have full fragile X syndrome mutations and no abnormalities in the PWS region. Fragile X analysis is widely available and should be considered in cases of childhood obesity.

#### **Obesity linked to Chromosomes**

There are also several well-defined chromosomal abnormalities that are associated with increased risk for obesity. Individuals with Down, Klinefelter, and Turner syndromes have a high risk of becoming overweight and obese.

**Down syndrome** is caused by three copies of chromosome 21 and is a highly recognized genetic syndrome. Individuals with Down syndrome can develop obesity stemming from environmental causes such as overeating and lack of exercise.

However, several features of Down syndrome can also explain a higher prevalence of obesity. Individuals with Down syndrome can have short stature, hypotonia and hypothyroidism, all of which increase the likelihood of becoming obese.

**Klinefelter syndrome** occurs in males and is also known as XXY syndrome. Characteristics of this syndrome

*Hainline continued on page 10*





## Just A Survivor

*By Janalee Heinemann*

Recently, I received an e-mail from Velma, who volunteers with the One List. She and I were dealing with some misperceptions about some of the members of PWSA (USA). Velma said that many of the younger parents were in awe of me and "tend to forget that you are a parent too, flesh and bone, emotions and feeling." This statement made me stop and reflect on how time and situations can change perceptions.

I reflected back to my first impressions of the board at the first conference I attended. I remember being a little intimidated by them and feeling that they came across as a clique that I could not approach. Little did I know that they were simply parents and professionals caught up in the business of the association who were once in my shoes – and now I am in theirs. As Marcel Proust once said, "The opinions which we hold of one another...are in no sense permanent...but are as eternally fluid as the sea itself."

That night, I did some reflecting and soul searching, and would like to share with all of our members excerpts from my response to Velma.

I had not stopped to realize that there is a new generation of parents who do not know me as a parent first, and executive director of this organization second.

Most of the older generation raised their children side by side with me. We laughed and cried and fought the outside world together. Their respect came not because I was on a pedestal – but because through my writings and our phone calls, they knew I was in the trenches with them. (A wise professor of mine once told me, "Don't let anyone put you on a pedestal, because sooner or later, they will knock you off it.") I was the first parent to write honestly about a PWS parent's fears, grief, anger and embarrassments. It is easy to write about our pride and triumphs – but in between those great moments in life, there are many more humbling and frightful hours.

It makes me sad to think that at some point other parents stopped looking into my heart and started looking, as Velma stated, "in awe" of me. The truth is I am simply a survivor like the rest of you on the One List.

**I survived...** a yearlong, bedfast, serious illness as a child and the death of my dad at an early age.

**I survived...** almost dying myself after the birth of my second child, and wondering if I was going to live through the birth of my third child, along with long nights of fear wondering who would raise my babies if I died.

**I survived...** the kidnapping of my first grandchild and the molesting of my daughter by a favorite uncle, with the subsequent guilt, fear, legal and safety issues.

**I survived...** a divorce after 17 years of marriage.

**I survived...** being very poor, and going back to high school while juggling the care of three toddlers and volunteer

work.

**I survived...** driving two hours one way to school at nights and on weekends to get my undergraduate degree while raising grade-school children, foster children and working full time as a child abuse worker.

**I survived...** threats on my life and the lives of my children for taking in black foster children in an all-white community.

**I survived...** the joy and fear of getting a full Dean's Fellowship to pay for my Master's degree – knowing I could only keep it if I got straight A's, and that I could not afford to stay in school without it.

**I survived...** All of this before PWS came into my life!

While getting my degree, I took a course on personal and interpersonal skills. During one session, my professor had us close our eyes and try to envision the animal we were and the animal we would like to be. I got a strong image (of all things) of me as a cockroach – and what I wanted to be was a butterfly.

In pondering why, I realized that the cockroach, with its thick shell of armor, is one of the longest living survivors. To become a butterfly and be free, I would have to take the risk of shedding my tough outer shell, and take some risks.

So I became a butterfly and spent the next 15 years with my heart and soul open while working with children with cancer and Hospice for 13 years – and those children did the same for me.

One of the things I love about working with people who are forced to look beyond this world is that they are at the most "real" time of their lives, and are children and adults of great substance.

During this time I was living with and writing about PWS. There was no support system in Missouri, there was no e-mail and very little had been written about it. We felt our only hope was to attend a national conference. I had quit my job to raise Sarah and Matt, and was in school studying for my Master's degree. We were struggling so much financially that we had to stay in a tent during that first conference – and we were not by any means campers! But, as always, we survived and thrived. The rest is history.

When I was asked to take on the job as executive director of PWSA (USA), I had real reservations. As one provider whom I respect said to me some time ago, "I work with many people who have children with disabilities, but I find parents of children with PWS to be the toughest to deal with. They are so angry!"

At the time, I was a bit insulted by that remark, but reflected on it and realized that there may be some truth in the statement. I think it's because we spend our life not only fighting the

*Director continued on page 15*





## May Our Hopes Become Reality

*By Lota Mitchell*

The International Prader-Willi syndrome conference of 2001 is one I shall never forget! It was the conference I went to — but didn't attend. It was the conference where I became PWSA (USA) president — in absentia. And it was the conference I had to leave to go to North Carolina because of the birth of my new grandson who couldn't wait for his Fourth of July due date.

Because the event precipitating my sudden departure was a joyous one, I can't feel too badly about missing the conference, although all the reports I've heard since then say it was outstanding. Thanks go to Jim and Joan Gardner for a terrific job!

One regret, though, was that I didn't get to see all the little ones, who are so adorable. At the early conferences the youngest children were probably 8 or 10. This year there were ten in the 0-1 age group and a total of 45 in the 2-5 group! There were few in those early days over the age of 30 because most died in their teens and twenties of obesity-related complications. Current conferences have many in their 40s. What a difference now! Thinking of this made me reflect on all the changes that have come about since the birth of my daughter, Julie, 31 years ago.

There was NO diagnosis when my beautiful baby girl was born. My father-in-law, a general practitioner before he went into internal medicine, had delivered more than 300 babies and had never seen one like her. Three very experienced and very puzzled pediatricians examined her, and none had a clue. For the next several years I swung like a distraught pendulum between hope and fear. Then, when Julie was five, I read a newspaper article about the first PWS clinic in the world in Seattle, Washington, and the words "Prader-Willi syndrome" entered my vocabulary — to stay.

I didn't want to believe my own diagnosis, but NO laboratory tests existed to confirm or disconfirm it. It was not until she was 19, when she participated in the definitive research on uniparental disomy, that we knew for sure. In her first half dozen years or so, there were NO publications for us to read or share with teachers and doctors, NO idea of what caused her condition, NO e-mail and NO other parents or anyone else to turn to for support, information or advice.

There was NO growth hormone at the time when she would have benefited from it, NO group homes although I knew she would need one when she became an adult, NO psychotropic medications to ease if not cure, NO genetic counseling with any meaning.

What a difference now! Today many, if not most, babies are being diagnosed early on, sometimes within days of their birth. If they live in areas where such help is available, they are receiving a variety of therapies to help them along their developmental way. PWSA(USA) offers a wealth of information through

literature and videos. Parents can network via chapters and e-mail. Growth hormone, for those who are able to take it, increases muscle mass, height and normal body configuration. For the adults, more dedicated group homes are springing up, as well as other creative housing alternatives, where they can feel they have a life of their own, even though a greater or lesser level of supervision is necessary.

What a debt of gratitude we owe the researchers who have patiently put together the bits and pieces to bring us to the point we are now. Research breakthroughs are seldom dramatic events that stand alone, much as we'd like to believe that, but rather are the result of many hours, months, even years, of painstaking, nitty gritty labor — like the bricks that, one by one, finally create a house. How exciting it was that this year's International Conference featured not one day of sharing of scientific research, but two and a half days as researchers from around the world came to report and network.

But is it enough? No! The most difficult challenges of Prader-Willi syndrome — the appetite leading to obesity and the behavior problems — have yet to be conquered. The effort is there. PWSA(USA) has had a research fund since 1983 which provides start up funding grants for projects which have been submitted to our Scientific Advisory Board (SAB) and recommended by the SAB for Board approval. In addition, PWSA (USA) provides support for research through our newly formed Research Clearinghouse Project, by playing a strong role in networking researchers (for example, the conference), and advocating for appropriate research.

But is it enough? No, not yet! The effort must move ever forward. PWSA(USA) — and all its members — must continue to fund research, and offer support, networking, encouragement and appreciation to all who help make a better life for our children.

I, like Martin Luther King, have a dream. I have a dream that answers will be found to control appetite, so that all those with PWS can eat normally, cease to suffer the relentless pangs of hunger, and my daughter can join us for the Thanksgiving holiday. I have a dream that answers will be found to control mood and behavior effectively, so that today's nightmares may become just bad dreams of the past, and those with PWS will be able to control themselves as least as well as the rest of us (who certainly aren't perfect!).

And I have the greatest dream of all — that the ultimate answers will be found, so that all with PWS can be "regular" children and adults (Julie's word for those who are "normal").

We all have a dream, the same dream. And we all have a hope — a hope that the dream will become reality, and soon.



# To All The 2001 Conference Volunteers

## If it weren't for you....

200 children and adults with Prader-Willi syndrome from 26 countries, 80 siblings, and 371 parents would not have experienced the joy and bonding that this conference brought to all.



▲ A group from PWSA (UK) with PWSA (USA)'s Janalee Heinemann



Snack break! ►

## If it weren't for you...

123 scientists presenting from 20 countries would not have been able to share their research with 130 scientists and medical professionals from all over the world.

## If it weren't for you...

a family from India would not have the knowledge and resources to go back and educate an entire nation.



◀ PWSA (USA) members join with a family from India

## 2001 World Conference Scholarship Contributors

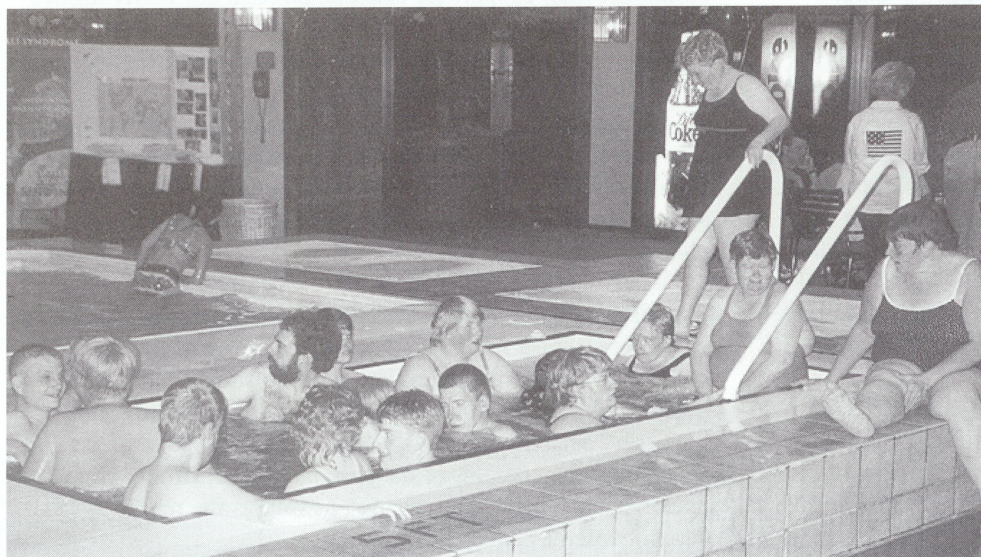
Minnesota Chapter  
Prader-Willi Syndrome Association (USA)

The Pittsburgh Children's Institute  
The Prader-Willi Foundation, Inc. of New York

PWSA of Indiana  
PWSA of Oklahoma  
William Capraro

Fran Cohen  
Pauline Haller  
Mary & Bob Hill  
Carolyn & Jim Loker  
Kenneth & Ruth Prettyman  
Cheri Clothier  
Whitney Olsen  
James & Jeanne Hanchett

37 families were registered at no charge (68 adults, 17 siblings, 18 with PWS); 25 families received hotel room grants; 4 received air fare; 3 received meal assistance; 3 received other travel assistance.



▲ Question: How many people with PWS can get into a hot tub?  
Answer: There's always room for more!



# and Sponsors

## If it weren't for you...

a mother from Japan might never have been able to meet the mother from the USA with whom she has corresponded for 4 years. Although words were not well understood, the hearts and smiles needed no words.



▲PWSA (USA) members join with new friends from Japan

## If it weren't for you...

we may not have realized that love has no borders and that together, we could "Fly High."

## If it weren't for you...

international delegates from 25 countries and PWSA (USA) chapter presidents would not have had an opportunity to share their common issues and goals.

Bill Capraro and  
Stephanie Combs  
▶



We just want to congratulate everyone on a TREMENDOUS conference.... We are home with nothing but beautiful memories and mountains of notes. Our most sincere thanks to you all. What a great team!

*Vodia and Ross Parker, Canadian PWS Org.*

## A World of Thanks!



◀  
Matt  
Heinemann  
and  
Anna Loker

## Thanks to you...

friendships bloomed  
heavy hearts were lightened  
lives will be saved



## A world of thanks to Joan and Jim Gardner

How do we thank the couple who volunteered 3 years of their lives to undertake the daunting task of coordinating the most complex conference on Prader-Willi in the world? Jim and Joan Gardner – we will be forever grateful. You did the impossible! For many years to come, the fruits of your dedication will impact the world of PWS throughout the USA, Australia, Austria, Belgium, Canada, Chile, Denmark, England (UK), Finland, France, Germany, India, Ireland, Israel, Italy, Japan, The Netherlands, Norway, Poland, South Africa, Spain, Sweden and Switzerland.



include hypogonadism, infertility, long limbs, progressive enlargement of the breast tissue, truncal obesity and increased risk for certain kinds of tumors.

Testosterone therapy has proven useful in patients with XXY to normalize adolescent development and increase muscle mass.

**Turner syndrome** occurs in females and is the usual result of losing one X chromosome during fetal development. Short stature, broad chest, developmental abnormalities of the heart and large blood vessels and neck webbing are common characteristics.

Growth hormone replacement therapy is becoming common treatment for short stature individuals with Turner syndrome.

### **Leptin deficiency and MC4R mutations**

Several genes have also been linked to severe childhood obesity without being associated with other congenital defects characteristic of traditional childhood obesity syndromes.

The leptin gene, a homolog of the mouse gene *ob*, has been mapped to chromosome 7. Leptin deficiency leading to morbid obesity has been recently described in two families. Leptin mutations have been associated with morbid obesity, increased appetite and hyperphagia, and poor or slow sexual development. Both of these families illustrated autosomal recessive inheritance.

Leptin replacement therapy has been initiated with success in patients with congenital leptin deficiency. Weight reduction was sustained due to decreased appetite and food consumption. Gonadotropin (brain sex hormones) levels also increased after 12 months of leptin therapy. This treatment may help prevent the lack of sexual development seen in adults with leptin deficiency.

Leptin receptor mutations have also been associated with the characteristics of leptin deficiency plus initial growth retardation and hypothyroidism due to changes in the hypothalamus of the brain.

Three sisters have been described with high serum leptin levels with mutations in the leptin receptor gene on chromosome 1. These individuals had normal birth weight, early growth defi-

ciency and hyperphagia that led to obesity. Autosomal recessive inheritance was suggested.

Another gene with a possibly greater effect on the presence of obesity in the general population is MC4R, located on chromosome 19. MC4R, melanocortin receptor, is present in high levels in areas of the hypothalamus that are known to be involved in feeding behavior. Mutations in MC4R have been described in several families and have been found by screening populations of obese children and adults. Studies have found that 3-5 percent of individuals with a BMI greater than 40 have a mutation in the MC4R gene. Most individuals have one copy of the changed gene and one copy of the normal gene, implying autosomal dominant inheritance for obesity associated with MC4R mutations. MC4R mutations appear to cause a "pure" obesity syndrome; no other endocrine abnormalities have been associated with mutations.

### **Head trauma may be a factor in obesity**

A thorough evaluation of obesity in children should also include consideration of head trauma affecting the hypothalamus. This condition is termed hypothalamic obesity and is thought to arise from overeating due to poor regulation of satiety and hunger.

Besides obesity, hypothalamic insult can lead to other endocrine abnormalities including growth hormone deficiency and hypothyroidism, both of which exacerbate obesity. However, hormone replacement has not proven to be effective in treating obesity in these patients.

Hypothalamic obesity can be seen in children with brain tumors, those who have undergone surgery, or those who have had radiation. One study of patients with craniopharyngioma showed that these patients have increased serum leptin concentrations and severe obesity. This is contrary to expected effects of increased leptin, based on obese patients with leptin deficiency. A proposed explanation is hypothalamic insensitivity to endogenous leptin due to the underlying brain damage of study patients.

### **Psychological impacts of obesity**

Obesity, regardless of cause, has

psychological impacts on affected individuals and families. In children who are mentally retarded, that impact may actually be greater on the parents and caregivers. Our society favors and holds individuals who are not overweight/obese in better esteem. This attitude is overtly expressed in the media, and a great amount of resources are aimed at persuading individuals that weight loss is not only desired, but also required for a successful life.

Many studies have shown that obese children demonstrate increased psychopathology and social problems when compared to non-obese peers. The data have also suggested that weight loss leads to improvement in psychological functioning. Many previous studies have used children seeking psychological or psychiatric treatment as a study population to observe differences between obese and non-obese children. It is very important that a correct diagnosis be made because of the varied treatment options available that are syndrome specific. Quality of life can be improved through treatments, and better psychological adjustment can be achieved through proper family education about a particular diagnosis.

*References available on request*

## **NIH mailing sent**

We recently participated with the National Institute of Health in a mailing to inform the membership of a research study that we are supporting and partially funding.

We received a few phone calls from concerned members about the release of our membership mailing list.

Please be reassured that all envelopes were addressed here at the PWSA (USA) national office. We did not release a membership mailing list. NIH provided the envelopes and performed the mailing in order to minimize the mailing expenses.

We apologize for any confusion that this may have caused.

*Peggy Fox, Business Manager*



# Abstracts from the Scientific Sessions

Co-Chair Suzanne B. Cassidy, University of California, Irvine, USA

Co-Chair Daniel J. Driscoll, University of Florida, Gainesville, USA

Conference Administrator Cathy H. Smith, University of Florida, Gainesville, USA

**Genetics** Moderator: Arabella Smith, Royal Alexandra Hospital for Children, Westmead, Australia

Speaker: Robert D. Nicholls, University of Pennsylvania, USA

**Genome Organization, Function and Imprinting in PWS.** Robert D. Nicholls,

Jinghua Chai, John M. Greally, Devin P. Locke, Todd A. Gray, Jessica Knepper, Joan H.M. Knoll, Evan E. Eichler, Shinji Saitoh, Daniel J. Driscoll, Yin L. Ge, Satya P. Kalra, Dabney K. Johnson, Eugene M. Rinchik and Tohru Ohta

Speaker: Karin Buiting, Institut für Humangenetik, Essen, Germany

**Imprinting Establishment and Maintenance in the Prader-Willi/Angelman Domain on Human Chromosome 15.** Karin Buiting, Maren Runte and Bernhard Horsthemke

Speaker: David Ledbetter, University of Chicago, Illinois, USA

**Proximal 15q: Physical Mapping and Implications for Diagnostics and Mechanisms of Rearrangements.** D.H. Ledbetter, S.K. Mewborn, J.A. Fantes, C.M. Lese, J. Hedrick, R. Brown and S.L. Christian

**Reactivation of Imprinted Genes in PW S.** Shinji Saitoh, Kyoko Takano, Maki Takahashi, Akira Sudo and Takahito Wada

**Pitfalls in the Diagnosis of the PWS: Differential Diagnosis with other Chromosomal Aberrations.** Annick Vogels, Marie-Jozef Descheemaeker, Veerle Govers and Jean-Pierre Fryns

## **Obesity and Metabolism**

Moderator: Merlin G. Butler, University of Missouri-Kansas City, USA

Speaker: Satya Kalra, University of Florida, Gainesville, USA **Genes and Hypothalamic Pathways in the Control of Obesity and Metabolism.**

Speaker: Tony Holland, University of Cambridge, Cambridge **Investigating Brain Function and Behaviour in PWS.**

Tony Holland, Joyce Whittington, Jill Butler, David Clarke, Harm Boer and Tessa Webb.

**Open-Label Pilot Study of Topiramate in Adults with PWS.**

Nathan A. Shapira, Mary C. Lessig, Wayne K. Goodman, Helen C. McCune and Daniel J. Driscoll

**Early Diagnosis of PWS and Implications for Nutritional Management.** Margaret S.N. Gellatly

## **Medical/Dental**

Moderator: Daniel J. Driscoll, University of Florida, Gainesville USA

Speaker: Daniel J. Driscoll, University of Florida, Gainesville,

**Medical Issues in Adults with PWS.** Daniel J. Driscoll, Brian M. Smith, Janet H. Silverstein, Arlan L. Rosenbloom, Douglas W. Theriaque and Alan Hutson

**Cortical Excitability in PWS: A Transcranial Magnetic Stimulation Study.** Carlo Cividari, Graziano Grugni, Paola Naldi, Roberta Vicentini, Roberto Cantello and Roberto Mutani

**Young Age at Demise Among Persons with PWS.** Arabella Smith, Georgina Loughnan and Stewart Einfeld

**Speech in PWS: Fluency, Voice and Articulation.** Truus Defloor, John Van Borsel and Leopold M.G. Curfs

## Dedicated to the Memory of Professor Andrea Prader 1919 - 2001

**A Child with PWS and Ethylmalonic**

**Aciduria.** Merlin G. Butler, Majed Dasouki, Susan Hunter, Niels Gregersen, Ali Naini and Salvatore DiMauro

## **Animal Models**

Moderator: Suzanne B. Cassidy, University of California, Irvine, USA

Speaker: Arthur L. Beaudet, Baylor College of Medicine, Houston, Texas, USA

**Models of PWS and Genetic Analysis of *Snrpn* in the Mouse.**

Arthur L. Beaudet, Jan Bressler, Ken-Shiung Chen, Ting-Fen Tsai, Mei-Yi Wu, Monica J. Justice, Yong-hui Jiang and Carlos Bacino

Speaker: Camilynn I. Brannan, University of Florida, Gainesville

**Mouse Models of PWS.** Camilynn I. Brannan and Stormy J. Chamberlain

**Regulation of *Snrpn* Expression in PWS.** Stormy J. Chamberlain and Camilynn I. Brannan

## **Growth Hormone**

Moderator: Martin Ritzén, Karolinska Institute, Stockholm, Sweden

Speaker: Ann-Christin Lindgren, Karolinska Institute & Pharmacia Corporation, Stockholm, Sweden

**Improved Growth and Sustained Body Composition During Seven Years of Growth Hormone Treatment in Children with PWS.**

A.C. Lindgren and E.M. Ritzén

Speaker: Aaron Carrel, University of Wisconsin, Madison

**Beyond Height: Metabolic Effects of Growth Hormone in Children with PWS.** Aaron L. Carrel, Susan E. Myers, Barbara Y. Whitman and David B. Allen.

Speaker: Marja Thorén, Karolinska Institute, Stockholm, Sweden **Effects of Growth Hormone (GH) Therapy in Adults with PWS.** Charlotte Höybye and Marja Thorén

**Beneficial Effect of Growth Hormone Treatment in Children with PWS Less Than Five Years of Age: A Retrospective Analysis.** Linda Bone Jeng, Shauna Heeger, William T. Dahms and Suzanne B. Cassidy

**Satisfactory Effect of Growth Hormone on Height in Patients with PWS.** Toshiro Nagai, Satoru Sakazume, Atunori Yoshino, Kazuo Obata, Kyoung-Chang Kim, Nobuo Murakami, Takayasu Murai and Ryoichi Sakuta

*Abstracts continued on page 12*

## Abstract Booklets Available

For a limited time, prior to their publication in a professional journal, Abstracts of the papers presented at the 2001 PWSA/IPWSO Conference will be available from the PWSA (USA) office in Sarasota, Florida.

The cost is \$25.00. Those wishing to purchase copies should contact the PWSA (USA) office by telephone at 1-800-926-4797, or by e-mail at pwsausa.org.



### Endocrine (Non-Growth Hormone)

*Moderator:* Susanne Blichfeldt, Rigshospitalet, Copenhagen, Denmark

*Speaker:* Anthony Goldstone, St. Bartholomew's Hosp., London, UK  
**Hypothalamic Neuropeptides in PWS.** Anthony P. Goldstone, Unga A. Unmehopa, Stephen R. Bloom and Dick F. Swaab

### Gonadal Failure in Adolescents and Young Adults with PWS.

Susanne Blichfeldt, Liselotte Skov, Niels Illum and Jørn Müller

### Fertility in Women with PWS: Two Case Reports.

**Case 1. A Woman with PWS gives Birth to a Child with Angelman Syndrome.** Astrid Schulze, Helle Mogensen, John R. Østergaard and Karen Brøndum-Nielsen.

**Case 2: A Woman with PWS gives Birth to a Healthy Baby.** Arne Åkefeldt, Carl-Johan Törnhege and Christopher Gillberg

### Psychology and Psychiatry

*Moderator:* Leopold Curfs, Clinical Genetics Center, Maastricht, The Netherlands

*Speaker:* Elisabeth Dykens, University of California, Los Angeles USA  
**Are Jigsaw Puzzle Skills "Spared" in People with PWS?**

Elisabeth Dykens

*Speaker:* Leopold Curfs, Clinical Genetics Center, Maastricht, The Netherlands

**Event-Related Brain Potentials in PWS: An Evaluation of Attention and Inhibition in the Visual and Auditory Modality.** Leopold Curfs, Marjo Brinkman and Johannes E.A. Stauder

**Object Alternation Performance, Eye Blink Rate, and Obsessive-Compulsive Behaviors in PWS: Implications for Brain Mechanisms.** Laura Holsen and Travis Thompson

**Long-Term Follow-up of Children with PWS: Are Some PWS Children More Vulnerable for Developing Psychotic Problems Than Others?** Annick Vogels, Marie-Jozef Descheemaeker, Veerle Govers and Jean Pierre Fryns

**Findings from a Population-Based Study of PWS.** Tony Holland, Joyce Whittington, Tessa Webb, Jill Butler, Harm Boer and David Clarke

**Childhood Rituals and Compulsive Behavior in Children with PWS.** Anastasia Dimitropoulos and Travis Thompson

**PWS and Autistic Spectrum Disorder: A Chance or an "Intimate" Relationship.** Barbara Y. Whitman

**Compulsive Behavior in PWS: Possible GABAergic Mechanisms.** Travis Thompson, Merlin Butler and Anastasia Dimitropoulos

**The Lessons of PWS and Chromosome 15: 1. Is There a Heuristic Value in Differentiating Psychiatric Diagnoses? 2. Psychiatric Symptom Clusters in PWS and their Heuristic Value in Implications for Further Research** Judith A. Brice, Barbara Whitman and Jeanne M. Hanchett

**Attitudes of Siblings Towards Having a Brother or Sister with PWS.** Erica L. Burner, Shauna Heeger, Jane Robinson, Matthew Warman, Evan Eichler, Suzanne B. Cassidy and Anne L. Matthews

**Neuropsychological PWS: The Special Case of Mathematical Abilities.** Laura Bertella, Stefano Marchi, Enrico Molinari, Graziano Grugni and Carlo Semenza

**Self-Injurious Behavior in Prader-Willi.** Palma Bregani, Irune Achutegui, Laura Bosio, Laura Deni, Isabella Coppa, Giuseppe Chiumello

**PWS due to 15q11-q13 Deletion in a (13;14) Robertsonian Translocation Carrier.** Fanny Cortés, Bianca Curotto, Lorena Santa María and Angélica Alliende

**A Polymorphism at the Diagnostic SNRPN Not I Site Produces**

**Atypical Methylation Patterns in Analyses for PWS.** Jean M. DeMarchi, Scott Chartrand, Karen Treat and Bernice A. Allitto

**Description of 60 PWS Persons: Cross Sectional and Longitudinal Data.** Mie-Jef Descheemaeker, Veerle Govers, Annick Vogels, Jean-Pierre Fryns

**The Educational Support to Keep Compliance with Domestic Injection of Growth Hormone (GH) in PWS.** Kumiko Egawa, Michiko Hara, Yumiko Sasaki, Fumiko Fukazu, Tetsuro Yamanishi, Tomoichi Imaizumi and Katrina Reyes

**Relationship Between Parental Attitudes and Behavior Characteristics of People with PWS.** Mai Funahashi, Tokuzo Harada, Mie Murakami, Michiyo Azumi and Kana Hattori

**Unsuspected Obesity Hypoventilation Causing Cardiopulmonary Deterioration in Children with PWS.** Linda M. Gourash, James E. Hanchett and Jeanne M. Hanchett

**Communication Skills and Social Interactions in Adolescents and Adults with PWS.** Veerle Govers, Mie-Jef Descheemaeker, Annick Vogels and Jean-Pierre Fryns

**Retrospective Review of Development and Behavior of Monozygotic Twins with PWS.** Louise Greenswag, Don C. Van Dyke, Jane Matzen, Deborah Downey, Dennis Harper, Anne Tabor, Sherri Smith and Eva Tsalikian

**PWS: A Developmental Perspective.** William Griffith

**Resting Metabolic Rate in PWS.** Graziano Grugni, Gabriele Guzzaloni, Giuliana Mazzilli, Dario Moro and Francesco Morabito

**Patient with PWS and Oculocutaneous Albinism.** Engela M. Honey, Arnold L. Christianson and Elizabeth J. van Rensburg

**NDN and MAGEL2 as Candidate Genes for PWS.** Syann Lee, Jocelyn M. Bischof, Sharee Kuny and Rachel Wevrick

**One Year Follow-up of Five PWS Children Diagnosed During the First Month of Life.** Pierre Moulin, Véronique Delagnes, Maïte Tauber, Corine Alberge, Marie-Claude Bloom, Anne-Cécile Perier, Joëlle Tricoire and Michel Rolland

**Diagnostic Investigations in the Group of 77 PWS Polish Patients.** Ewa Obersztyń, Agnieszka Szepecht-Potocka, Ewa Bocian, Jerzy Bal and Tadeusz Mazurczak

**Ventriloquism for Communication in Patients with Mental Retardation (MR).** Yumiko Sasaki, Michiko Hara, Yoko Sakurai and Kumiko Egawa

**The Incidence of PWS in Australia: An APSU Study.** Arabella Smith, J. Egan, E. Haan, G. Warne, P. Montgomery, C. Oley, G. Ridley, E. Elliott and K. Williams

**A Cost-Effective Protocol to Exclude an Imprinting Mutation in PWS and Angelman Syndrome.** Arabella Smith, Luke St. Heaps and Lisa Robson

**Ethical Aspects of Prevention of Obesity in PWS.** R.H. van Hooren, G.A.M. Widdershoven, M.J.J.M. Candel, H.W. van den Borne and L.M.G. Curfs

**Body Composition in PWS Compared with Nonsyndromal Obesity: Relationship to Physical Activity and Growth Hormone Function.** Edgar G.A.H. van Mil, Constance T.R.M. Schrandt-Stumpel, Leopold M.G. Curfs, Klaas R. Westertep, Willem J.M. Gerver, Wouter D. van Marken Lichtenbelt, Arnold D.M. Kester, Guido A.K. Heindendal and Wim H.M. Saris

**PWS and Cycloid Psychosis.** W.M.A. Verhoeven, S. Tuinier and L.M.G. Curfs  
*Ed. Note:* Because of space limitations, references to "Prader-Willi Syndrome" have been abbreviated to PWS.



# A Hurrah for Aaron

By *Steve Carvajal*

Aaron Carvajal (pronounced carve-a-hall) was born on March 1, 1998. Born six weeks premature, extremely weak, and unable to breathe without the help of a ventilator, Aaron was off to an extremely rough start. My wife Susan and I nevertheless had high hopes for our firstborn son. Tiny, yet beautiful, our perfect son was going to be okay once he recovered from the affects of the anesthesia they had given Susan when they did the C-section. The doctors assured us that after two or three days he would perk up.

Three days passed, then a week, and still he showed no signs of growing stronger or more alert. The doctors began to worry, and MRIs followed CAT scans, and still there was no answer. But obviously something was wrong with him. He would not wake up unless he was picked up, rubbed and moved and talked to, or bathed. The minute he was put back down, his eyes would flutter shut, and he would be asleep once again. At least your average baby is able to cry if something is wrong, but with our son we never heard a sound. Aaron would not wake up from hunger, nor would he wake up from a soiled diaper. He literally slept 23 hours a day.

Finally, one doctor requested a geneticist to check on him. At first Aaron was misdiagnosed with an extremely rare genetic disorder known as Angelman's Syndrome. We were told that he would never speak, would never mentally mature past the age of 18 months, and would not walk till he was at least 8 or 10 years old. The little shred of hope that he still might be OK had now slipped from my fingers.

The geneticist decided to be thorough and run further blood tests to confirm his findings. We were later told that rather than Angelman's, Aaron instead had Prader-Willi: a slightly less rare and less debilitating syndrome. Aaron would one day speak, and Aaron would walk sooner, but he would need help. Aaron would need extreme physical and occupational therapy to help him reach his full potential. This would begin as soon as we were able to take him home from the hospital. A month and a half after he was born, Aaron came home.

Our lives, which had been on hold while our son had drowsed the time away in the pediatric intensive care unit (PICU), now had to resume. I must give full credit to Susan for all that happened to bring Aaron to where he is now. I was a basket case. I was drowning in depression, and barely able to get out of bed in the morning to go to work, and could not have been able to see to the needs of my utterly helpless son. Susan got on the phone and arranged for therapy. She got on the internet and found out all she could about the syndrome, its treatment, and any financial assistance that might possibly be able to help us with our staggering medical bills: six weeks in the PICU, plus hundreds of hours of physical, occupational, and speech therapy, running on average 100 dollars an hour.

Time passed, and at 1 year we celebrated that Aaron was strong enough to finally roll over. At 18 months Aaron became strong enough to sit up. By the time he was 2, he had progressed



*Aaron Carvajal rides on Blackjack*

to the point where he was able to stand while holding on to furniture. Twice a week, Aaron's physical therapist would work with him, and tell us that very soon he would be able to walk, and yet walking eluded

him. Frustrated by his lack of progress, Susan, having read about the benefits of therapeutic horse back riding, was able to get in touch with Kerstin Fosdick of the Saddle Light Center for Therapeutic Horsemanship. We went one Thursday evening to watch a session, and had mixed emotions about it.

How could we put our tiny, fragile, handicapped son on so huge an animal as a horse? But we prayed and discussed, and argued, and finally decided to give it a try. Aaron never had a moment's doubt. With the help of the instructor, Linda Koehler, and three volunteers (two to walk on either side of the horse and hold onto Aaron, and another to lead the horse), he took to riding as if it were as natural as breathing. There was a confidence in his eyes when he rode upon Blackjack that I had never seen there before. He rode straight upright and with an ease that I could never have imagined, much less believed if I had not seen it with my own eyes.

When Aaron began to walk in September 2000, we were stunned. Shakily, he would pull himself up to a stand, and then launch off, taking two or three steps before falling to the ground. That was truly the happiest day of my life.

After that, I felt certain that the horseback riding had really made a difference. I had thought of it as a fun activity that would build his confidence — and that was plenty — but after that I truly began to notice the time and thought that Linda gave to his sessions. I noticed the increasing difficulty of the stretches and the maneuvers. I noticed Linda's concern and dedication.

It is now April 2001, and on the 7th we participated in the Saddle Light walk-athon. Aaron walked almost half of the course with no help whatsoever. Yes! The son that doctors had told us may not walk till he was 6 or 7 years old, walked almost a whole mile at the age of 3! I cannot describe the feelings of joy and gratitude that I have associated with that day.

Aaron will never be able to grow up, move away and have a family of his own. But he is an extremely happy and loving child. He will continue to progress at his own pace and I will thank God for every milestone that he reaches, thank God for every smile he gives me when he wakes up in the morning, and thank God for every step he takes to escape me when I tell him it's time to take his medicine. I don't know how long it would have taken him to learn to walk without Saddle Light's assistance, but I do know that he has progressed further than I would ever have imagined he could.

*The Carvajals are from San Antonio, Texas.*



# Daughters are Special People

By Teresa Kellerman

The house is quiet on this Sunday morning as the boys sleep in and I read my latest copy of *The Gathered View*. Reading stories other parents have written about their children always renews in me my love for my own special kids. (All my kids are “special kids.”)

The coffee is ready and I open the cupboard to choose among my many coffee mugs. I choose according to my mood, and this morning I am in a daughter mood as I look forward to my Sunday chat with my Mom later today. I choose the mug that says “Daughters are Special People.” Yes, we are! There’s a special bond between a mother and daughter. It’s a bond I share with Karie, although our relationship is very different from the relationship between my Mother and myself, but both are based on compassionate love and patient understanding.

As I pour my coffee, I recall my visit with Karie a few days ago. Since Karie has PWS, I have to be careful on her visits with me. I have to be sure the alarm is set on the refrigerator and that the pantry is locked because of her insatiable appetite. I choose to lock up the food to help relieve her anxiety about getting into food that might endanger her health. I have to be prepared for one of Karie’s “moods.” I never know when she will have a “Prader-Willi moment,” when she is not in control of her emotions.

This is not always a bad thing, as she can be uncontrollably happy as well as grumpy or angry. When she is in happy mode, she might laugh and squeal with delight and talk non-stop about the delicious meal that her group home staff prepared the previous evening. When she is grumpy, it is not by choice, and there is not much I can do to change her mood, but I try to accommodate her emotional state, whatever it might be.

On this particular visit, she was in a relatively good mood. She helped me choose clothing to pack for our upcoming trip to Minneapolis for the International PWS conference. She was so agreeable to accepting choices I had made when shopping for new clothes for her.

Karie did not choose to have PWS. Nor did she choose to have the array of frustrating disorders that come with PWS genetics — insatiable appetite, slow metabolism, various obsessions and compulsions and stubbornness. Because of her special diet, I could not offer her goodies to eat, like I might do with other family members or friends who come to visit. So I offered her a choice of a can of diet soda or a cup of tea. She chose tea. I lifted two coffee mugs out of the cupboard and offered her a choice.

Now those of you who understand PWS know that choices are important (I usually limit it to a choice of two or three to

simplify the choice-making process), and that quantity is more important than quality. Most folks with PWS will pick the bigger portion or item when given a choice. The two mugs were of different sizes and the bigger one had a picture of candy canes

on the front. This is Karie’s mug, and I know she really likes that one. The smaller mug was my “Daughters are Special People” mug. I asked her which one she wanted me to use for her tea, and she said, “Daughters are Special.” She looked at me and gave me one of those smiles that a parent never forgets. The look said, “I love you, Mom, and I like being your Special Daughter.” Of course, that moment will be cherished in my memory forever.

Karie’s choice meant a lot to me for several reasons. First, she has not been especially loving to me lately, because I have had to be in “Mean Mom” mode - enforcing a stricter diet for her

after a drastic weight gain in the past few months, and having to make other tough decisions as her guardian about which she has not been too pleased.

Also, my role as mother has been shared by several other women in the past year as her group home staff have deepened their relationship with Karie. These six women who provide care in her home give so much more than is required by their job description; they give her the same quality of care that I gave her when she was still living with me.

So her choice of the smaller mug and the telling look she gave me were especially endearing gestures. She acknowledged me as her mother, and acknowledged the love she gets from me as her Special Daughter.

Today I will prepare the registration forms for Karie and me to attend the PWS conference, and in the weeks ahead I will prepare myself for a week one-on-one with Karie. I expect this will be a combination of grumpiness and glee, filled with all sorts of “Prader-Willi moments.” On the way home from the conference, Karie and I will take a short one-day side trip visit to my parents’ home, where we will enjoy a three-generation reunion of parents, grandparents and daughters.

My Mom and Dad chose to be the best parents they could be. My Mom took parenting courses and read books and magazines on parenting. Even with 10 children, she managed to make each of us feel special and loved. In parenting Karie, I follow my parents’ example and choose to be the best parent I can be, and I am a strong advocate for her. I have been called “tenacious,” and the term suits me well. I don’t give up until Karie gets what she needs to be safe, healthy and happy. I don’t have to be tenacious with my Mom and Dad regarding Karie’s needs. Her Nana and



*Special Daughter Karie and Teresa, her mom*

*Special continued on page 15*



## Survivor - continued from page 6

syndrome, but also fighting all of the prejudices and lack of understanding that constantly surround us. The problem is, we do not know when to take off our armor and boxing gloves.

So, Velma, I can understand that the One List is where you would want people to feel safe and be able to fly free before they have to put on their own personal coat of armor and face the rest of the world.

Velma, I know you must look at all of the years ahead and wonder if you can be strong enough handle it all. Yes, you can. None of us was born strong. We learned how to be strong because we had no choice.

I agree that we all need to remember to take off our armor and put down our boxing gloves when dealing with each other. We have enough battles to fight. Everyone is chopping at the tree

– not realizing that they are chopping down the very tree that we are all sitting in.

It is the tree that is saving all of us from the raging rivers of the “early years” of the syndrome when there was no hope or help, and protecting us from the inhumanity of the world beyond PWS. Is it a perfect tree? **No**. Does it need fertilizing and shaping? **Yes**. But each of us is a leaf or branch or a twig on that tree, and each of us has a role to play and a choice to make as to whether the tree dies or survives – and our children with it.

Thank you for reaching out your hand. What keeps me going is knowing that underneath all of the armor and the anger, each and every parent I have met is really a beautiful and caring person... and a survivor just as I am.

## Special - continued from page 14

Poppa show their love by sending Karie cards and notes, and when we visit, Nana plans nutritious and tasty meals that meet Karie's nutritional needs. And she always gets loving hugs, regardless of the mood Karie might be in when we visit.

The joy I felt the other day when Karie came to visit me was a reflection of the love she feels as my Special Daughter and the commitment of care from her other mothers. The joy I feel today is a combination of peace at knowing my Sweetie is loved, and in knowing that I am loved as a Special Daughter too. Because of the difficult nature of PWS, I have mixed feelings about taking this trip with Karie, but I do look forward to the short time I will get to spend with my daughter and my parents together. Karie can't always choose to be in a cheerful mood, but I can choose to do whatever I can to make this conference and the visit with her Nana and Poppa enjoyable.

As I finished writing this story, I received two phone calls, almost simultaneously: one from Karie and one from my Mother. Karie was having a crisis and needed her mom's intervention. She was having a bad day. Not by choice, but by the nature of PWS, and her inability to accept an agreement that was made the day before. My mother called because she was just thinking about me, and chose to pick up the phone and connect with me. After 3 or 4 more phone calls from Karie, I think I solved her problem. After talking this morning with my mom, and sharing all the joys and frustrations of dealing with Karie lately, I'm feeling replenished with mom energy, and I'm also feeling deep appreciation for my mom's support in helping me cope with parenting a daughter with PWS. Mothers are special people too!

Now my coffee mug is empty, but my heart is full of tenderness for my Sweetie, and of gratitude for my own mom, who taught me all about being a good mother and how wonderful it is to be a Special Daughter.

*Teresa Kellerman is the President of Prader-Willi Syndrome Arizona Association and the mother of Karie, 26, who has PWS.*

### The Chuckle Corner

Our 3-year-old daughter, Brenna (who has PWS) likes routine. After 10 a.m. Mass on Sundays we usually go to get bagels at Ronnie's. Brenna loves plain mini bagels.

On this particular Sunday, we had other plans, and did not have time to go to the bagel store. Brenna asked for her “mini” and we informed her that we weren't going to Ronnie's.

Not taking no for an answer, her reply back was, “so what about a Mickey?”

*Mary Walsh  
River Dale, New Jersey*

*Do you have a joke or funny story to share with readers? Please send your stories, including your name, address and phone number or e-mail address, to the PWSA office.*



## PW

## POST OFFICE

For people with PWS



Dear PWSA

Little kids, it is not so bad to go group home. You need to follow directions. The people or staff will let you go places like zoo, restaurant, go to store. People who run the house, you follow their direction. They will go with you to do things. Who's know, you might get girlfriend, or boyfriend when you at group homes. Let's talk about Matt Heinemann, well I will live with my girlfriend with her mom. In the future I will marry Lori Kimley. She my baby doll, sweet girl. She is the nicest girl I meet on my job. She really care about me.

*Matt Heinemann  
St. Louis, Missouri*



# Acknowledgements

*Our Sincere Thanks for Contributions Received in June and July*

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*Due to the volume of memorial contributions for  
William Capraro Sr., Frank Moss and Tara Salvors,  
they will be listed in the next GV.*

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.

