



The *Gathered View*

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



Jose Rodriguez - Peru

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The Pennsylvania Mini-Conference: Learning New Strategies

By *Lota Mitchell, Associate Editor*

Daniel Opp, 9 months, the "Little Professor" from Lancaster, Pa., and Ashley Tate, 10 months, the "Little Princess" from Westchester, Pa., were among the many participants at the mini-conference November 6 sponsored by the Prader-Willi Syndrome Association of Pennsylvania. They were highly popular, with much "oo"-ing and "ah"-ing and "coo"-ing by their numerous admirers, who flocked around them.

The chapter was proud to present the full day event, held in Carlisle in central Pennsylvania. Featured speakers were Dr. Linda Gourash, previously medical director of the Prader-Willi Syndrome/Behavioral Disorders Program at the Children's Institute in Pittsburgh, and Dr. Janice Forster, who was director of psychiatry with the program. They had been very popular and outstanding presenters at the national conference in July.

The exciting news is that their presentations will be available on DVD. Deep gratitude and thanks go to the California Foundation, the Pennsylvania and the Minnesota Chapters, Oakwood PW, Inc. of MN, and an anonymous donor for their grants making the DVD possible. All the wonderful information Drs. Gourash and Forster had to impart, addressing issues for both children and adults, will be available through PWSA(USA) and IPWSO when all the necessary editing is completed.

Titus Keiningham, the videographer who has worked with us in the past, arrived on the scene the day before, took a look at the room where the program was to be held and promptly went out to buy some beautiful deep blue fabric to hang as a backdrop.

Tots Ashley Tate, the "Little Princess," (top) and Daniel Opp, the "Little Professor," (center), who have PWS; conference organizers and presenters Pam Eisen, Linda Gourash, Maria Silva, Lota Mitchell and Janice Forster (bottom)



Mini-Conference continued on page 13

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

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

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

Adults With Prader-Willi Syndrome PWSA (USA) Advisory Board Meeting

Sharing Insights About Having Prader-Willi Syndrome

On October 22 and 23, 2004, members of the Adults with PWS Advisory Board held their annual meeting at Saw Mill Creek Resort in Sandusky, Ohio. As you can imagine, the days were filled with excited conversations and profound insights.

On the first full day of meetings, Advisory Board members participated in an interactive group session. This facilitated exercise served to draw out experiences and feelings related to having PWS. Several objectives were met, including providing support to the adults involved, openly discussing daily challenges and solutions in a safe environment, and instilling

What do you want others to know about what it is like to live with PWS?

Mike: How hard it is to control the diet • How hard it is to exercise and stay fit and trim • That we need to have friends • How hard it is not to steal food • How hard it is to control our temper

Brooke: How to help us with food issues • That we have close family relationships • How hard it is to manage weight • How important it is for us to exercise • To understand our behavior issues – that sometimes we can't help it

Shawn: • How hard it is to learn how not to eat everything set before you • How hard it is to accept help in food control • Shawn's Tips: Help us make good choices like giving us the option of having fewer calories at breakfast and lunch so we can have more at a special dinner • Staff should come into our home with a good attitude and not yelling at us • Always keep the cabinets and pantry locked up and also the refrigerator and freezer locked with a chain and locking key

Diane: • The very, very strong urge to eat even it means stealing food and risking going to jail to eat • Constant need to eat, eat, eat • How difficult it is to keep from exploding (behavior) over food

Abbott: • How hard it is being hungry all the time • How hard it is to lose weight • How hard it is to cope with PWS – getting up every morning and wishing you did not have the syndrome – and trying hard to accept yourself • What it is like to be picked on all the time • How hard it is not to steal food

Goals and challenges...

What Makes You Happy?

Being at my goal weight • Working out, being healthy and alive • Doing my own laundry and checkbook • Making friends, having hobbies and social activities • Being in my own apartment • Independence – but I know I cannot handle it • Having my own car – but that is also a problem •

confidence in these young adults as role models and leaders of others who have PWS.

The second day of meetings continued with the themes of day one — conversational turn-taking, mutual support and leadership — then transitioned into a session of art therapy. Members made thoughtful greeting cards, in hopes of mass production for use as an ongoing Advisory Board fund-raiser. I thank all who participated and now share their insightful thoughts and feelings with our members.

— **Mary K Ziccardi,**
Board Advisor

Adults With PWS Advisory Board members: front row, L-R: Shawn Cooper, Mike Nacht, Diane Arnoudse, Brooke Fuller, Abbott Philson; back row, Advisors David Wyatt, Janalee Heinemann, Mary K Ziccardi



To have a job – not be so isolated • Having my cats • Spending time with my family • To advocate for myself and others – ARC board, etc.

What are the issues in your life you want control of?

My temper, behavior – getting aggravated, annoyed • Stealing • What I eat, my weight • No property destruction, no verbal aggression • Sneaking food • My money – locked up so I do not spend it wrong • Stuck-ness – transitions are hard • More control over food – I think I can, but I need help

What are the issues in your life you want others to give you more control of?

How to take care of my animals – some don't respect that I may know more what to do • Choosing my own staff • Staff having a car to take me around • Have support staff knock before they come in

What Is Missing/Needed In My Life?

Missing loved ones who have passed on • Wish my brother lived close • More freedom • My own apartment • Special college, school • A more challenging job — being with people • Chemical for us so we don't feel hungry • More group homes

PWS Advisory Board continued on page 12



Our Crisis Committee Responds ASAP

Carolyn Loker

I would like to highlight the volunteer work of PWSA(USA) board member Mary K Ziccardi. We are very fortunate to have Mary K on the board, now serving in her 4th year. She is an administrator of REM OHIO, Inc., a residential provider for people with developmental disabilities.

The Crisis Committee, chaired by Mary K, is another important committee sponsored by PWSA(USA). Other members include Ken Smith, vice president of PWSA(USA); David Wyatt, crisis counselor; and Janalee Heinemann, executive director. The purpose of the Crisis Committee is to assist in decisions about allocating designated funding to aid people in crisis needing services such as travel grants to the nearest approved hospital or rehabilitation center. The committee also assists in

the review and approval of conference grants given to families.

Mary K also backs David Wyatt in handling crisis situations when he is unavailable or inundated. A recent letter to the national office stated: "I was so thankful to be in contact with Mary Ziccardi. What an outstanding lady! She was such a help to me, and she showed compassion and understanding."

In 2002 Mary K was instrumental in implementing the Adults with PWS Advisory Board, because she and PWSA (USA) believe that all people with PWS should have a voice. Mary K has been a strong advocate for people with PWS for more than a decade. It is often thanks to her that their voices and needs are heard.



Mary K Ziccardi

Paul Alterman and grandson Macao (without PWS) Paul's son Andy, the baby's uncle, has PWS

PWS Crises and The Heroes Who Help

By Janalee Heinemann

It is a new world for PWS, and although many of our young children are looking great and functioning extremely well, we cannot afford to ignore our families who have no where else to turn. Yes, more research is needed, and we now have programs for prevention and early intervention, but all too often we still get the call from a desperate parent. If not PWSA(USA), who will help the hundreds of families who have a child with PWS in crisis? Just looking at a few of the calls this week, I have to ask, if we were not here, who would assist...

...the 6-year-old who was suspended from school for "aggressive behavior" when the teacher took a French fry from his lunch to "teach" him to control what she thought was obsessive-compulsive behavior?

...the 8-year-old who weighs 200 pounds and whose behavior is out of control?

...the 21-year old in ICU at 385 pounds on a ventilator?

...the divorced mother who has cancer that has metastasized and who is trying to find placement for her 15-year-old daughter if she can no longer care for her?

...the mother of the young man who died suddenly and the medical examiner was refusing to do an autopsy which would give some answers about why he died?

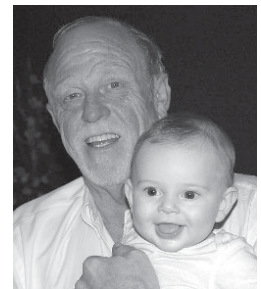
The Altermans and the Willetts

Two very important grants have made a world of difference in the lives of our families in crises. Since 2001, the Alterman Family Foundations have supported our part-time crisis counselor, David Wyatt. Many of you know what a crucial difference David's support has made for our

families. Over this time, he has worked with more than 1,900 PWS families in crises — from school, legal, behavior, medical, placement and abuse to advocacy for services. (I assist with serious medical crises.) As one mother recently wrote: "David Wyatt has been my grip on sanity more times than I can count, and (PWSA-USA) never charged me a dime. With no spouse or living family, David has pulled my 'fat out of the fire' numerous times and helped me to keep on keepin' on in many difficult situations with my daughter."

Thanks to the Willett Family, we've had an expansion in our Crisis Intervention and Prevention Program. Following is David's report of what we have been able to do with their generous donation.

"We have also been very fortunate in being able to expand our crises services thanks to another 'angel' family, the Willetts, who in honor of their nephew Brian Schertz, have supported a grant to be used for legal and educational crisis for the last two years. Through this support, we have been able to develop an educational consultant program. This program has been reserved for serious educational crises, where the problems were leading to suspensions, threatened expulsion, legal action, or location in another school that usually was for individuals with delinquent behavior.



Crises continued on next page

27th PWSA(USA) National Conference

July 28 – July 29, 2005 Sheraton World Resort, Orlando, FL

www.pwsausa.org Click on Conferences/Events

Plan NOW for 2005 Conference grant requests.

Look at alternative, local grant sources. Have your request in writing, be prepared before you call. Do not wait, many funds get used up early in the year.

- State Developmental Disability Council 1-800-695-0285 NICHCY or check their web site www.nichcy.org
- Check for funding through your local church
- Local Service clubs (Kiwanis, BPOE, etc.)
- The ARC (disability related funding)
- Parent-to-Parent (disability-related funding)
- Make A Wish (call Diane first before making application 1-800-926-4797)

And then...PWSA(USA) Grants. Making conference grant requests from PWSA (USA): Download Grant Application from our website www.pwsausa.org or e-mail: national@pwsausa.org to request a form; or call our 800 number to have an application mailed.

All grant recipients will be required to be or become members of PWSA(USA). If the membership cost is a problem, a Scholarship Membership can be requested.

So that we can assist more families, grants may not always include air fare.

All reservations for grant families will be made by the national office through our travel group.

Submission Deadline is April 30, 2005 for PWSA USA grant applications.

- E-mail grant requests to: national@pwsausa.org
- Fax to 941-312-0142
- Mail to:
Prader-Willi Syndrome Association (USA)
5700 Midnight Pass Rd. Ste. 6
Sarasota, FL 34242
Attn: Grant Committee

Crises - continued from page 4

“The initial situation for this program often involves attorneys for the school and the parents, crisis letters and information about PWS from the PWSA National office, and numerous telephone calls. Dr. Barbara (B.J.) Goff, Mary K Ziccardi and others have offered their expertise as educational consultants. After talking with all parties, they typically go to the school and help to develop a behavioral plan that is included in a new IEP. This intervention has laid litigation to rest and provided a model for the school and family. Most importantly, it has given the student with PWS a fresh start toward completing his or her education.

“As of November 2004, our specialists have consulted in South Carolina, Maine, Kansas, Florida, New Hampshire, Ohio, Missouri, Arkansas, Massachusetts, Nevada and California. In these 12 states there have been 19 consultations. Several more requests and exploration of the various possibilities have begun. Typically the cost is shared between PWSA (USA) and the school system.

“What makes their intervention successful? These consultants provide expertise in developing behavior management plans written into an IEP. They also develop valuable support for the school, teacher and family. They’ve confirmed that education for a child with PWS is really what most people involved want. None has been easy, but what they have accomplished is nothing short of miraculous!

“In the last year we have been able to use the Willett funding to also address some of the more serious legal and placement crises through educating staff. There are often legal charges against a person with the syndrome and the threat of expulsion from placement. Our consultants have

conducted training programs for the agencies in crises that provide the services for our young adults.

“One example was training sessions in northern and southern California by Dr. B.J. Goff. The session in San Diego, with more than 80 attendees, was video-taped and will be available for distribution in early 2005 to others with assisted-living facilities for people with PWS. The northern California session was a part of the state chapter meeting, where B.J. was able to train staff as well as talk to the parents of children with PWS about school behavior issues. This collaboration between the California Foundation and PWSA(USA) was made possible by special grant funding.

“Although the Willett funding has also been used for individual cases of serious legal and other forms of crises, the educational consultant program and the staff training program probably have the greatest impact for servicing a large number of people and impacting change.

“Our Crisis Intervention Program has grown each year. In 2003, I worked with 496 crisis situations and the numbers continue to increase. The year 2004 has been no different. In November alone, for instance, we received 55 requests for crisis services.”

Heroes come in many forms. Those identifying and living with the need — such as parents, providers and educators. Those with the skill to help — such as Mary K, B.J., and many others who volunteer their expertise to help our children. Those who can help fund the services needed. Support from two families — the Altermans and the Willetts — means hundreds of lives are rescued. There are many unsung heroes in our world of PWS. Let me be the first to sing the highest praise to our crises heroes.



Executive Director's View

Accelerating Into The Future Scientific Roundtable Discussion Weekend

Janalee Heinemann

PWSA(USA) and our Research Advocacy Team, chaired by Jim Kane, sponsored and coordinated a two-day November meeting in Baltimore where some of the finest minds in PWS research discussed the issue of hyperphagia (uncontrollable appetite). The theme was “Accelerate the Future,” defined by Jim Kane as “bringing to fruition today that progress which otherwise might be a long time coming.”

The weekend was an opportunity to share current knowledge, discuss ways to move forward in understanding the issues, identify crucial questions in hyperphagia-related research, and plan for new research on hyperphagia.

Researchers attended from the USA, United Kingdom and The Netherlands. All have specific experience in appetite control and metabolic problems with diverse backgrounds in genetics, molecular genetics, endocrinology, neurobiology, neuro-imaging, gastroenterology, psychiatry, and nutrition. A representative from the National Institutes of Health (NIH) also attended.

The resulting Strategic Plan for Research, to be presented to the PWSA(USA) Board in January, focuses the energy of PWSA(USA) using a new model and a narrow focus to make progress on PWS research at a pace never before seen. The new model emphasizes more collaboration among the scientific group, sources of funding and PWSA. Jim believes the narrow focus will put the right people in the right places with the right support to work on cutting-edge questions.

Here are some of the issues that I think you will find interesting.

* **Why a focus on hyperphagia?**

- Because it is the controlling force in PWS
- It is a causative factor in aberrant behavior
- It prevents our children from becoming independent
- PWS is a window into the “House of Obesity”
- There is commercial and federal potential in funding

* Exploring the **phases of PWS** identified more than the two phases traditionally reported. Children with PWS often become overweight at 1 ½ to 3 years even without an increase in calories — before the hyperphagia sets in, which can start from 3-12 years. Some adults with PWS who are 30-40 years old have been reported to have a reduction in the tremendous drive to eat. Furthermore, problems which manifest themselves in the newborn infant as “failure to thrive” most likely begin months prior to actual birth. The changes seen in early childhood — from failure to thrive, to normalcy, to thriving too well, to the raging appetite — are

intriguing to scientists, presenting many opportunities for study.

* **Diet:** Adults with PWS typically must be on 800-1200 calories per day, unless on growth hormone, and thus need multivitamin and mineral supplements. They also need adequate fluids and fiber to avoid constipation, due to weak muscle tone. We need to know how to treat diet better if the person has been on growth hormone. What has GH done to the energy-balance equation?

* Curiously, there are fewer instances of type 2 **diabetes** than expected for weight and less high fat levels in the blood than expected. Why?

* Many families are intrigued by **surgical options for treatment** of severe obesity, from gastric bypass surgery to experimental use of electric stimulators in the stomach area. There has been limited investigation into their application in people with PWS. We hope to learn from bypass surgery being done in Italy and are considering ways to instigate projects in the U.S.

* Abnormalities in **the brain**, particularly the hypothalamus, may lead to increased appetite in PWS, such as fewer oxytocin neurons (that inhibit eating) in PWS than controls. We need a more centralized approach and collaboration for the use and distribution of brain tissue for research since it is so difficult to get. Deep brain stimulation is being tried in some other brain diseases — the problem is where to position such stimulation in the brain of those with PWS.

* Genetic research has identified two major causes of PWS, deletion (types I and II) and uniparental disomy. Each one has variations such as in behavior and visual memory. What **variations** exist **between the different genetic types** in appetite and metabolic problems?

* **Drugs:** Ghrelin is the only known gut hormone that increases appetite, but high ghrelin levels in PWS are not the whole story. It is unclear whether low levels of other gut and pancreatic hormones that lower appetite contribute to increased appetite in PWS. These hormones can be given to patients or lowered by other drugs (e.g. intra-nasal PYY3-36, somatostatin analogues to lower ghrelin levels) to try and treat the appetite in PWS. However, the PWS brain may not respond normally. Would other drugs in varying degrees of development to treat obesity in the general population (e.g. those affecting the neuropeptide Y, melanocortin and other brain receptors; drugs to increase metabolism to burn off energy), be effective in PWS?

* The European Union (14 countries) recently commissioned establishing a centralized **database** project for people

Accelerating continued on page 14

Special Siblings Give Us A Different Perspective

Jeremy Eisen and
his sister
Gabriella,
who has PWS

By *Jeremy Eisen*

I have a younger sister with PWS. When Gabriella was born, her doctors said that she would not live; she lived. They said she would never walk; she walked. She never gave up.

At the time, the diagnosis was given as an unknown neuromuscular disease. Seven years later, she was correctly diagnosed with PWS, which is associated with diminished mental faculty, delayed motor development, short stature, and hypogonadism. The defining symptom, however, is the lack of satiety after eating. Coupled with an extremely slow metabolism, PWS is a recipe for obesity. Without intervention, it is a recipe for an early death.

Growing up with Gabriella was a challenge. I helped my father to install locks on the kitchen doors, the kitchen cabinets, and even the refrigerator. Although she scored only a 40 on the Stanford-Binet Intelligence Quotient, Gabriella always found ingenious ways to sneak food. What she lacked in raw intelligence she made up for with sheer determination. She never gave up.

Gabriella and I live on the same planet, but we live in different worlds. And yet, sometimes I cannot help but see that she and I are not so different. Sometimes, when I feel burdened by one of life's many vicissitudes, I think about my sister and the seemingly overwhelming obstacles she has had to fight — my troubles suddenly seem petty and trivial by comparison, and I feel inspired by her constant perseverance.

When I was in elementary school, children used to make fun of each other by using the word “special” in reference to



special-ed; children can be so cruel. Having Gabriella as my little sister, I have come to understand the true meaning of “special.” It is not simply the *nom du jour* of the politically correct, but a precise adjective. Having her in my life has enriched my life in ways that a normally functioning sibling could not.

Most people, when hearing of PWS, react with sadness. They want to know why. As a scientist, I know the mechanisms that cause PWS and I know that it happens to about 1 child in 15,000. But that is not the answer they are looking for when they ask why. To that, I answer not as a scientist but as a human being — I believe children like Gabriella are special because they compel the rest of us to be the best human beings possible.

Jeremy Eisen, 32, and his sister Gabriella, 25, are the children of IPWSO President Pam Eisen. Jeremy holds a bachelors degree in physics from Dartmouth, works as an internet technology consultant, and is now applying to medical school with the intention of becoming an ophthalmologist.

Make Sure You Are Counted: Help Us Complete Our PWS Database!

PWSA(USA) currently has the largest database collection of information on children with Prader-Willi syndrome, but some of it needs updating and all data on families lacks essential information that could be crucial to improve the medical care of our children.

Board Member Barbara McManus is helping to improve our database by increasing the size and accuracy of the information.

You can find the form on our web site at www.pwsausa.org/population/ or e-mail us at national@pwsausa.org, or call the national office at 1-800-926-4797 and ask to have a form sent to you.

Everyone who completes a form will receive a copy of our new Medical Alert booklet shown here, which is essential for doctor and emergency room visits.



In a perfect world, PWSA(USA) would have the names and pertinent information needed for everyone with the syndrome. There is power in numbers!

If we can acquire a more comprehensive database, National Institutes of Health (NIH) and other funding sources will be more willing to help our children through funding continued research in PWS and obesity. You don't need to be a member of PWSA(USA) to be included in this data collection.

You can help us help you, your family and all those who have been touched by Prader-Willi syndrome just by completing a form and becoming part of our national database. Please act now!

The Genetics of Prader-Willi Syndrome: *An Explanation for*

By Linda Keder

Part 1 of 2. Linda Keder, former editor of The Gathered View, originally published this article in the March-May 2000 issue. It was revised and updated in July 2004 with the assistance of Merlin G. Butler, M.D. Ph.D., who chairs the PWSA(USA) Scientific Advisory Board.

When the medical world first learned about Prader-Willi syndrome in 1956, doctors had no idea what caused people to have this collection of features and problems that we now know as PWS.

In 1981, Dr. David Ledbetter and his colleagues reported a first breakthrough discovery: Many people with PWS that they studied had the same segment of genes missing from one of their chromosomes. They had discovered the deletion on chromosome 15 that accounts for about 70% of the cases of PWS.

Since then, researchers have made a series of other important discoveries about the genes involved in PWS. Thanks to their perseverance, we now know much more about the several genetic forms of this complex disorder, and we have genetic tests that can confirm nearly every case.

Chromosomes and Genes: The Basics

To understand the genetics of PWS, it helps to have a basic understanding of chromosomes and genes. Chromosomes are tiny structures that are present in nearly every cell of our bodies. They are the packages of genes we inherit from our parents. Genes contain all the detailed instructions our bodies need to grow, develop, and function properly — our DNA.

Specific genes direct our cells to produce proteins, enzymes, and other essential substances. Each of our many genes is located on a specific chromosome. Most of our body's cells contain 46 chromosomes — 23 inherited from our mother and 23 from our father. (Egg and sperm cells normally contain just 23 chromosomes, because those are the cells that join in conception and provide the baby the right number of chromosomes.)

Twenty-two of the chromosome pairs are labeled with a number based

on their size (chromosome 1 is the largest pair, and chromosome 22 is nearly the smallest), and the two chromosomes in each numbered pair contain the same genes (one set from mother and one from father).

The changes that cause PWS occur on the pair known as chromosome 15. The 23rd chromosome pair is designated as the sex chromosome pair. This pair determines the baby's sex: XX for a girl, XY for a boy.

Changes or errors in genes and chromosomes are common in the formation of egg and sperm cells. Some of these genetic changes will have no effect when a baby is conceived; some will cause a miscarriage; and some, like those in PWS, will cause significant differences in how the baby develops and functions. While many genetic disorders are caused by a change in a single gene and can be passed down from parent to child, PWS is more complicated.

Genetic Characteristics of PWS

Some of the important genetic characteristics of PWS identified through research are:

- More than one gene is involved in PWS, and these genes are near each other in a small area of what is called the "long arm" of chromosome 15 — in a region labeled 15q11-q13. Scientists still don't know exactly how many genes and which specific ones are involved.
- The critical genes must come from the baby's father in order to function properly; the mother's genes in this area are "turned off" through a rare phenomenon called "genomic imprinting."
- There are at least three different chromosome errors that can keep these

key genes from working normally, and all result in the child having PWS.

The two most common errors that cause PWS can occur in any conception — in other words, PWS is not usually an inherited condition; it just happens. In very rare cases, however, parents may have a 50% chance of having another child with PWS.

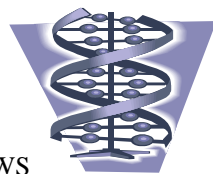
The Role of Genomic Imprinting

During the early 1980s, scientists puzzled over why some people who seemed to have PWS did not have the chromosome 15 deletion, and why some people with the chromosome 15 deletion seemed to have a different condition from PWS. Dr. Merlin Butler and colleagues began unraveling the puzzle when they reported in 1983 that the chromosome 15 deletion in PWS was on the father's chromosome.

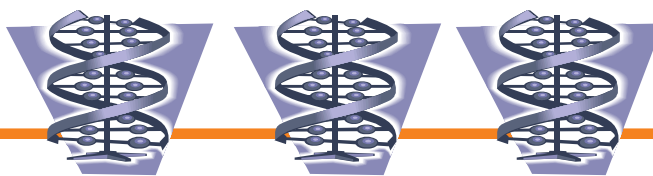
The next breakthrough came in 1989, when Dr. Robert Nicholls and fellow researchers announced their discovery that PWS is an example of genetic or genomic imprinting, a process well known in plant genetics but not previously identified in humans. This means that some of our genes have to come from a particular parent to work normally.

These rare genes are said to be "imprinted," or have the ability to be turned off or on, depending on which parent contributed the gene. In what scientists call the "Prader-Willi region" of chromosome 15 (the area where the deletion occurs), there are genes that must come from the baby's father that are active, or "expressed," in order to work. These genes are not active or expressed on the chromosome 15 inherited from the mother because the mother's imprint turns them off.

In Prader-Willi syndrome, these critical genes are either missing (deleted) from the father's chromosome



Genetics continued on next page



15, functioning improperly because of an imprinting defect, or the entire chromosome 15 from the father is missing and both chromosome 15's come from the mother. (See The Three Genetic Forms of PWS for more detail on each of these errors.)

When a deletion of chromosome 15q11-q13 region is found on the mother's chromosome 15, the result is an entirely different syndrome called Angelman syndrome (AS). That is because there is also one gene in the Prader-Willi region that is imprinted, or turned off, on the father's chromosome 15; people who lack this gene from their mother have AS rather than PWS. This discovery explained the mysterious cases of people who had a chromosome 15 deletion but did not have the characteristics of PWS — their deletion was on the chromosome 15 that came from the mother.

Because the genetic errors happen in the same section of chromosome 15, PWS and AS are sometimes called "sister" syndromes, even though the disorders have few features in common.

The Three Genetic Forms of PWS

Although every case of PWS is due to the baby failing to receive active genes from a specific section of the father's chromosome 15, there are three different ways that this can happen:

- Paternal deletion — about 70% of all cases of PWS

In the most common form of PWS, part of the chromosome 15 inherited from the child's father — the part containing the PWS critical genes — is missing. In some cases, the section that has disappeared (called a "deletion") is large enough to be identified in standard chromosome studies done with a microscope; in other cases, it is too small, but it can be detected with another chromosome test called FISH (see part 2, Tests Used To Diagnose PWS in the next issue of GV).

Genetic Subtypes in PWS

By Merlin G. Butler, M.D., Ph.D.

In 1981, a deletion of the proximal long arm of chromosome 15 was first described. Later in 1983, the deletion was reported on the chromosome 15 donated by the father.

The deletion of the 15q11-q13 region consists of about 6 million DNA base pairs from a total of approximately 3 billion base pairs found in all 46 human chromosomes. The chromosome 15 deletion is seen in about 70% of individuals with PWS.

This typical deletion is now recognized in two forms: Class (Type) I and Class (Type) II. There are three chromosome breakpoints (BP1, BP2 and BP3) located in the 15q11-q13 region — two breakpoints (BP1 and BP2) are proximally positioned close to the centromere while one breakpoint (BP3) is located at the distal end of the 15q11-q13 region (See Figure 1).

The Class (Type) I deletion involving breakpoints BP1 and BP3 is approximately 500 thousand DNA base pairs larger than the typical smaller Class (Type) II deletion involving breakpoints BP2 and BP3. PWS subjects with the larger Class (Type) I deletion generally have more severe behavioral problems. At least four genes are located between breakpoints BP1 and BP2 and clinical differences reported between PWS subjects with either the Class (Type) I or Class (Type) II deletion are apparently due to the loss of the four genes.

In addition to the PWS subjects with the typical paternal deletion of the 15q11-q13 region, about 25% of PWS subjects have maternal disomy 15 (both chromosome 15s from the mother). The observation of maternal disomy was first reported in 1989 and led to the discovery of genomic imprinting or the differential expression of genetic information depending on the parent of origin.

An imprinting center is located towards the middle of the 15q11-q13 region and consists of specific DNA which controls the activity of genes in this region depending on the source of the chromosome 15 which is inherited from the mother and the father. There are several genes recognized in the 15q11-q13 region which are active or expressed only on the father's chromosome 15 and under the control of the imprinting center. Hence, if these paternally expressed genes are deleted on the chromosome 15 from the father, or if both chromosome 15s come from the mother (i.e., maternal disomy 15), then PWS results in the child.

The two types of maternal disomy 15 in PWS are maternal isodisomy and maternal heterodisomy. Maternal isodisomy refers to the presence of identical genetic material (e.g., came from the same chromosome 15 in the production of the egg) and maternal heterodisomy which refers to the presence of genetic material from two different chromosome 15s from the mother. Children with PWS due to isodisomy may be at increased risk of having a second genetic condition if the mother is a carrier of a recessive condition such as Bloom syndrome.

Bloom syndrome is an autosomal recessive condition where the gene is located outside of the 15q11-q13 region but both members of the gene pair called alleles are abnormal. If the mother carries an abnormal gene allele on part of her chromosome 15s for this condition, she is unaffected because her other gene allele is normal (remember genes and chromosomes come in pairs). If that chromosome 15 region or segment containing the abnormal recessive gene allele for Bloom syndrome is donated in the egg by the mother to the child in an isodisomy

Usually a deletion happens for no known reason, and it is not likely to happen again in another pregnancy. There is nothing the father did (or did not do) to cause it and no way to prevent it. In rare cases of extremely tiny (micro) deletions, or when a chromosome change such as a “translocation” caused the PWS genes to be defective or lost, the family could have another child with the same condition. (In a translocation, part of one chromosome is broken off and attached to a different chromosome.) It is especially important for these families to have further testing and genetic counseling.

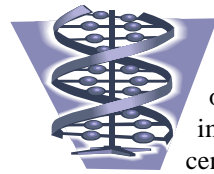
- Maternal uniparental disomy (UPD) — about 25% of cases

In this less common form of PWS, the baby inherits both copies of chromosome 15 from one parent — the mother. (Maternal means mother; uniparental means one parent; and disomy means two chromosome bodies). In these cases, the developing baby usually starts out with three copies of chromosome 15 (a condition called trisomy 15) because there was an extra chromosome 15 in the mother’s egg. Later, one of the three is lost — the chromosome 15 that came from the father’s sperm. The result has the same effect as a deletion. The child does not have active genes on chromosome 15 that must come from the father in order to be expressed (to function). Even though there are two complete copies of the mother’s chromosome 15, the key genes in the PWS region are imprinted, or turned off, in the mother’s copies. Because the error in this form of PWS starts with an extra chromosome in the mother’s egg, and older eggs are more likely to have errors of this type, older mothers are more likely than younger mothers to have a baby with this form of PWS. Even so, it is not likely to happen (and hasn’t yet) to a second child in the same family. When a baby inherits two identical chromosome 15s from the mother (isodisomy, or two copies of the same one rather than one of each of the mother’s own chromosomes), there is a

chance of having additional genetic problems or conditions.

- Imprinting defect — less than 5% of cases

In very rare cases, the PWS genes on the father’s chromosome are present but do not work because the imprinting process is faulty. The activity of the genes is controlled by a tiny imprinting center on chromosome 15 in the same area as the PWS critical genes. Normally, when genes are passed down to a child, the prior imprints are cleared away, and new imprints are made according to the sex of the parent.



When there is a microdeletion or other defect in the imprinting control center, gene function

on the father’s chromosome 15 may not be set to work normally. An imprinting defect can appear suddenly, or it can be present in the father’s chromosome that he received from his mother. If he received the defect from his mother, the father would not have PWS himself (because it’s on his maternal chromosome 15), but he could pass it on to his child (it would be the child’s paternal chromosome 15).

There is a 50-50 chance that any child he has will receive the chromosome with the defect instead of the one that’s working correctly. Likewise, the father’s siblings could carry and pass on the mutation to their children. More testing and genetic counseling are especially important for families who have a child with this form of PWS.

Subtypes - continued from page 9

fashion (i.e., both chromosome 15s contain the same identical genetic information or alleles) then PWS results as well as Bloom syndrome.

Less than 3% of PWS subjects have a defect of the imprinting center either in the form of a micro- or atypical deletion involving the imprinting center or due to the failure of the imprinting center to control the activity of genes in this region. The types of genetic errors (deletion or non-deletion) of the imprinting center are referred to as epimutations. A recent research study reported that in PWS subjects with imprinting defects that about 15% will show a micro- or atypical deletion of the imprinting center while the majority (85%) will have an imprinting defect but without an identifiable deletion using molecular genetic techniques. PWS subjects with an imprinting defect would have an abnormal genetic methylation testing result consistent with PWS; however, they have normal chromosome studies and DNA polymorphism studies utilizing parental DNA showing bi-parental or normal inheritance of both

chromosome 15s (i.e., one intact chromosome 15 inherited from the mother and one intact chromosome 15 from the father). Parents of PWS children with an imprinting defect may be at an increased risk of having additional children with PWS. Therefore, genetic counseling should be undertaken to determine the most accurate recurrence risks.

Thus, several genetic causes and subtypes for PWS do exist but all involve the chromosome 15, specifically the 15q11-q13 region. A further explanation of the genetics of Prader-Willi syndrome with illustrations can be found at the PWSA(USA) website.

Dr. Butler chairs the PWSA (USA) Scientific Advisory Board. Correspondence should be sent to him at: Children’s Mercy Hospitals and Clinics, Section of Medical Genetics and Molecular Medicine 2401 Gillham Road Kansas City, MO 64108. Direct phone: (816) 234-3290 Fax: (816) 346-1378; e-mail: mgbutler@cmh.edu

Fund-raising



Le Masquerade Nets \$95,000+ for PWSA(USA)

Thanks to Bill and Tina Capraro of Oak Brook, Illinois, Le Masquerade, the elegant dinner dance and live auction they hosted to benefit PWSA(USA), raised a net donation of more than \$95,000.

Two years ago we wrote that their last fund-raising event was “the largest fund-raiser ever hosted by a PWS family in the 27-year history of PWSA(USA).”

They have now far surpassed their own record!

This gala evening of fun and masks was in honor of their daughter Lea and was underwritten by Cimco Communications, Alan Bocca Jeweler, Jane & Maria, Ltd., Source 4, and VR Creative Services. A big thank you goes to all of the Capraros’ wonderful friends and relatives who attended the event and donated so generously.

Bill and Tina’s goal was to get PWSA(USA) “out of the red” for the year. Thanks to an overwhelming show of support through this and other special events (to be reported in the next edition) at the end of 2004, we have made it into the “black.”

As I noted in our crisis report on page 4 of this issue, “Heroes come in many forms.”

— *Janalee Heinemann*

Golf Tournaments Score Big for PWS

By Lota Mitchell, Associate Editor

What’s lots of fun, friendly, out in the fresh air, competitive — and raises money for PWSA(USA) and its chapters? Of course — a golf tournament!

Several of our chapters do annual golf outings, such as Pennsylvania and New Jersey. Individuals, too, have sponsored golf outings, like Bill Dunn, a neighbor of Clint Hurdle’s, whose daughter Madison has PWS, and Ira and Ronnie Levine, whose granddaughter Josilyn has PWS. Pat Meakim, whose daughter Bridget has PWS, was permitted to use the name of the company where he is employed, Cal-Chip Electronics, to encourage his customers to participate in his highly successful first attempt at putting one on. He plans to do it again in 2005.

These events have raised anywhere from a couple thousand up to as much as \$30,000, so it is a project well worth undertaking.

Hosting a tournament can actually be a great experience, too. Bill Vucci, father of Maria who has PWS, tells his inspiring story below. While each situation is different, Bill has a lot of good suggestions about putting one together.

If an individual or a chapter is interested in possibly sponsoring one but is uncertain how to go about it, the national office has a good list of experts, like Pennsylvania’s Mike Fabio and Mike Azzara, who have “been there, done that” and can give direction. National office also has sample flyers. Ann Coyne, national’s financial manager, notes,

Lea Capraro (R) and her good friend Anna Loker (L), who have PWS, enjoyed dressing up for Le Masquerade; Lea’s parents, Tina and Bill Capraro (bottom), helped get PWSA(USA) “into the black” for the year.



“This could a wonderful annual source of income for PWSA (USA), the guys and gals love it, and everyone has a good time.” Ann can answer questions about legal matters such as 501(c)(3) non-profit organizations.

Bill Vucci’s Golf Tournament

Last January, my wife Anne and I decided to host our first golf tournament. We knew nothing about golf, except that this sport is one of the most popular fund-raising events around the country. All we had was a lot of enthusiasm and passion to generate interest in others for PWS. We knew enough about PWS based on our youngest daughter, Maria Christine, as well as the quality information from national and our local chapter.

We envisioned an open tournament where anyone could play. Since many in our community knew about our daughter, we called it “The Prader-Willi Open — In Honor of Maria Christine Vucci.” I asked relatives, friends, chapter families and PWSA(USA) for ideas about running an event.

In February-March, I checked out various golf courses and chose one whose manager was genuinely interested in helping our cause. The green fees were reasonable, which created a larger profit return on our fund-raising efforts. I began asking local business owners if they could sponsor a golf tournament to help save our children’s lives. We were

Golf continued on page 13

Residential Supports...

Do you have what you need to be successful? If not, what is missing?

Staffing • Small refrigerator to put pre-prepared food in that I can have access to on a daily basis • Treadmill and other exercise equipment • My own phone line • More freedom from parents • Given more of my own control by my parents • My own decision making

What qualities should your group home staff have that will help you most?

No smoking • Energetic young females that want to work in a unique living situation • To help me make good choices – food and exercise wise • Patience, being kind to me • Great personality – not too old and has some spunk left, tolerance, great attitude • Someone who will talk with me, exercise with me, go on outings with me, and can drive me around • Being respectful – being treated like how you would like to be treated – knock on my bedroom door before coming in

What is the worst quality in a staff member?

Leaving the kitchen unlocked • Lazy, on the phone all the time • Lying • Not being confidential about me • Abusiveness, yelling • Stupid – don't know what is going on and unable to communicate with me what I need to know • Unsupportive • Giving me too much to eat on my plate • Not helping and encouraging me in my daily routine

What actions should cause a staff to be fired or removed from working with you?

Sexual or physical abuse • Laziness, lying • not supportive • Not helping me follow my schedule – not keeping me safe from food

Vocational Supports...

Successful? – What helps? What hurts?

Good supervisors and job coach • Successful – someone watches the food and what I get • Unsuccessful at work setting – a refrigerator there that was too tempting • Successful – been there 5 years – can't go in the break room or would lose my job – I take orders and help customers with their problems – I have sign on my desk: "Don't give me food or money because my job is rewarding enough." • Don't allow candy jars at work • If I get a bonus or gift card, they keep it and give it to my job coach. Direct deposit of checks so I am not tempted to spend it on food.

Parent and Family Issues...

What do you enjoy most about spending time with your family?

Vacation, joint outings together and having parties • Getting out of my apartment and doing things with them • Having the pantry locked with the key – it makes me feel more secure • Spending time with my grandfather • Watching the deer in the back yard • Having someone to vent to • Going to the movies, for coffee, or dinner

What part of your relationship with your family is most difficult?

They leave too much food around that tempts me • My parents are always asking me if I am losing weight, or ask if I am trying hard enough • My dad does not take criticism well himself on things he expects from me (e.g. healthy eating) • Spending too much time with one grandma who is critical of me • My sister is not tolerant – she does not want anything to do with me because I have PWS – she is a teen. (Another explained how her sister used to be that way, but grew up and "got over it") • My parents blamed me for their divorce

Is there someone in your family who "doesn't get it" about you having PWS?

Aunt and uncle – they have things I should not eat when I visit – too high in calories • Dad's two sisters – they put no food restrictions on me when I visit • My sister

What steps can we take, as a group, to help them understand?

Put them on the Dr. Phil Show • Have media coverage at the conference meeting • Publish information from this meeting • Speak as a group at conference

If I had one wish...

To have a normal body – but not lose the gifts I do have • I wish others could feel how we feel – know how we (all with PWS) have to live day by day • That people could have two days that they have to live in our shoes • It would be to not have the syndrome at all

Our thanks to the Gerald J. and Dorothy R. Friedman New York Foundation for sponsoring this meeting through a special grant.

The Chuckle Corner

Waste Not!

My son, David (age 12, who has PWS) has always been very interested in learning (and telling) about all the naughty things he or his family members have done as children.

A couple of years ago, I was telling him about a time when my mother was about 5 years old. She was washing dishes with her older brother, and got so angry with him that she cracked a plate over his head.

David looked at me with a horrified expression, amazed (I thought) that his gentle grandmother would have done such a thing. All became clear, though, when he looked at me for reassurance and said, "She finished the food first, didn't she?"

Carol Hearn, Plymouth, Minnesota

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



Golf - continued from page 11

pleasantly surprised at the positive response local businesses gave us. It taught us that when you have a child directly affected, you bring the passion for your cause that generates enthusiasm from others. Businesses are continually inundated with donation requests, so you need that strong personal interest to inspire their collaboration in your efforts.

In April-May, most of the preliminary work was done. Location, fees, sponsorship levels, team positions, competition games, dining plans, brochures and field refreshments were planned, but we had no committee yet and the event was scheduled for August 27, only 3 months away.

Since Anne is a well known teacher in our local parish, we started there. On June 5 we hosted our first Prader-Willi Open committee party at our house. It was a family event, and we bought lots of refreshments and even rented a clown to entertain everyone's kids as parents reviewed our plans.

Never in our wildest dreams did we expect the support we received! We gave out our plans, including sponsorship levels, timelines and information booklets about PWS and why it was important to bring awareness as well as fund-raise to support our national association. This helped everyone to answer questions when approaching potential participants. We stressed that every penny was going toward PWSA(USA), a 501(c)(3) non-profit organization. By the end of this meeting, every parent was interested in helping our cause. Positions, projects and timelines were in motion and everybody knew their jobs.

We called our team "The Fun Bunch" for good reason. From June on, we had a combined party/progress meeting weekly at our house. Our treasurer obtained a post office box for registrations and donations. Our secretary noted progress, new ideas, sponsors and work to be done. The prizes/awards team brought information.



The fearsome foursome of Chris Riley, Matt Hepler, Tournament Director Pat Meakim and Keith McCrone played in the Cal-Chip Golf Tournament for PWSA(USA). Pat's company, Cal Chip Electronics, helped sponsor his first golf fund-raising event.

We used a 501(c) (3) tax ID stamp and Xeroxed every check before sending to national so we could send personal thank you's in addition to the formal response from PWSA (USA). This will ensure future support for another event.

I worked on media events to bring in participation and donations. Radio stations interviewed us. Television stations hosted our family, and one station played the interview throughout tournament day.

Our original goal was to bring awareness of PWS to thousands of families and raise \$25,000, but we surpassed that! We earned \$28,000 and received media calls regarding further awareness interviews. Golfers, The Fun Bunch and businesses all want to be involved again. The Prader-Willi Open was a complete success!

Mini-Conference - continued from page 1

Soon chapter members were hauling into the room greenery from the lobby, the swimming pool, or wherever they could find it, and the hotel covered the audience tables with deep rose cloths. What a transformation!

Next morning, while babies, children and adults with PWS and their siblings enjoyed their own activities, more than 90 adults, many of whom were providers of residential care, filled the room to hear the presentations. The doctors primarily addressed behavior and its management, certainly one of the most, if not *the* most, pressing issues of those who deal with individuals of all ages with PWS.

Dr. Gourash, a developmental pediatrician with a life-long interest in education, discussed food, behavior and beyond, starting with basic nutritional facts and moving on to managing weight and behavior together. Then she outlined some new and groundbreaking skills and strategies for parents and caregivers, based on the No Doubt, No Hope principles. (Curious about that? Be sure to get your own

copy of the DVD. Watch for notice in *The Gathered View* when the DVD becomes available.)

Neuropsychiatrist Dr. Forster described "normal" Prader-Willi behavior and then Prader-Willi "Plus", when behavior goes beyond what is expected of the syndrome. Then she addressed the big question of whether to medicate or not, including the goals of psychiatric treatment, alternative coping skills, informed consent, and other issues related to the use of psychotropic medications.

Evening brought the banquet for young and old together, followed by a D.J. Before long, just about everybody, from baby Daniel dancing with his mom Joyce to Dr. Forster and Dr. Gourash, was out on the dance floor doing the Chicken, the Macarena, and shaking it up to "My Name's Not Willy!"

Underneath orange hats for everybody, lots of dinner calories got burned, and a good time was had by all.

We Remember...

Every person has something special to offer this world — and we, along with their families, want to share who they were and what they meant to the people who loved them.

Tragic Fire Devastates the Plotke Family

Early in the morning of November 30 we had a terrible house fire. My 15-year-old daughter Raizy was quick to get 8-year-old Jochanan, who has PWS, and 3-year-old Hadassah out of the house.

Kalman, age 11, was asleep in his bedroom with his door locked. The fire had started in the room next to Kalman's. I was unable to rouse Kalman to full wakefulness before the smoke was too thick. The result was that we lost our treasured, darling son Kalman to smoke inhalation. Our



Kalman Plotke

sorrow simply cannot be described. Kalman was very creative, a musician, he played guitar. He had already designed his CD covers and named his songs. He also took wonderful photographs. He was very funny and kept us all laughing. This past Thanksgiving he said, "I am thankful for my family, and for

my parents who love me and I love them, and they try to do what is best for me and they do what is best for me."

Accelerating - continued from page 6

with PWS. Several U.S. institutions are creating a database through a collaborative grant from NIH for rare disorders, a project involving the collection of extensive verified data on a limited number of individuals with PWS.

Meanwhile, PWSA(USA) is setting up an extensive database to collect **basic information on as many people with PWS as possible**. Each of these data collection efforts promises to build excellent sources of information on PWS for researchers.

One major problem is a serious lack of geneticists currently being trained. With early diagnosis, we have had a great insurgence of babies now diagnosed with PWS — but who is going to care for our children?

The Roundtable Weekend, the first such gathering tailored to a discussion of PWS and the raging appetite and metabolic factors, was considered a success by all attendees.

A longer report about this Acceleration weekend is available in the Members Only section of the PWSA(USA) web site or by calling 1-800-926-4797.

Our thanks to the Maryland Private Foundation for providing a portion of the funding for this Roundtable.

After Thanksgiving dinner we all sat down to play "Hear Me Out." Kalman loved to play board games. We all laughed so much and had a great time together.

The last thing Kalman was doing before he went to sleep on the night of the fire was to practice guitar. I will always remember him this way, dedicated to music, making us laugh, a source of the greatest happiness to our family.

Please remember everyone: Don't sleep in bedrooms with a lock door, keep your door unlocked. There should be smoke detectors in every bedroom and a fire escape plan for every room. Do not use foam mattresses on your beds. Please take care of yourselves.

Some of you may remember Kalman from past conferences. We last attended one in St. Paul, Minnesota. Besides being part of YAAP, Kalman also spent a lot of time riding the elevator and running up the stairs to the top floor. If anyone remembers him in particular I would love to hear your memories of him.

Chana Plotke and Family (from Florida before the fire)

NOTE: This family not only lost a precious son, they did not recover one item from the fire. Anyone wanting to help can send a donation to: The Plotke Fund — Bais Menachem Synagogue, c/o Steve Stokalo, 1111 NE 179th St, North Miami Beach, FL 33162
— *Janalee Heinemann*

PWSA(USA) Sponsors Research On Behavior & Development in PWS

Dr. Elisabeth Dykens' research team has relocated to Vanderbilt University's Kennedy Center and is currently studying behavior and development in younger and older people with PWS. This study aims to identify the factors related to behavior problems in PWS, including specific neurotransmitters, age and genetic subtype. Findings will guide effective pharmacological and behavioral treatments in the future. We are also studying relative strengths in people with PWS, including skill with jigsaw puzzles and certain personality strengths and how these contribute to successful life outcomes.

We are looking for people with Prader-Willi syndrome aged 4 years through adulthood, and their families, to participate at the Kennedy Center at Vanderbilt University in Nashville, Tennessee. Study activities include a day of behavioral and neurochemistry assessments. All participants will receive a written report of study findings by the research team. For further information call Elizabeth Roof, Research Coordinator at 615-343-3330. Limited funds for travel for subject participation are available. This grant is sponsored by PWSA(USA).

— *Janalee Heinemann, Executive Director*

Heavenly Angel (\$500-\$999)

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PWSA of Florida
Ole & Kristin Dam
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King County, WA
Brevard County
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Metropolitan Chicago
Victoria Protsman



Correction

In the November/December 2004 issue of *The Gathered View*, we mistakenly identified the people in a photo on page 10. Shown with Sen. Arlen Specter were Michelle Shirk and her son Benjamin of Reinhold, Pa.

We sincerely regret the error.

In Honor Of

Siena Mehta
Nishat Mehta
Minesh & Nisha Mehta
**Mr. & Mrs. James Banks-50th
Anniversary**
Gilbert & Lorraine Marquez
Mike Burns - "teacher thank you"
John & Deborah Mosley-Duffy
Nicholas Busfield's 1st Birthday
Leanne & Keith Champagne
Joan & Leonard Ott
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A Conf. Session for Single Parents?

Program plans for the July 28-29, 2005 National Conference in Orlando, Fla. are now under way. Should we have at least one time slot focused on learning more about the needs and concerns of our single parents? Please tell us whether you, as a single parent, believe this would be helpful and supportive.

Please call or e-mail me your ideas about this at 1-800-926-4797 or cic@pwsausa.org. Are you in favor of such a session or not? Do you have other options to suggest?

— David Wyatt, *Crisis Intervention Counselor*

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