May-June 2001
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# The Gathered View

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

*Back to the Future*

# Our Road Trip To PWSA Conference

***By Janalee Heinemann***

*The following is an article I ran across that I wrote 13 years ago. Matt just turned 28, Sarah is 26, and our "little" grandson Mikey (who now goes by Mike to all but grandma) is in college. People and situations change, but the commonality of family dynamics remains the same. We thought anyone who has been to conference with children or plans to attend would be able to identify with our story. I hope to see you in Minnesota!*

It sounded like a great idea, at least something we should try once — the great family vacation. In our 7 years together, Al and I have always had the luxury of what we call "real" vacations. Most of them have revolved around the national PWS conference.

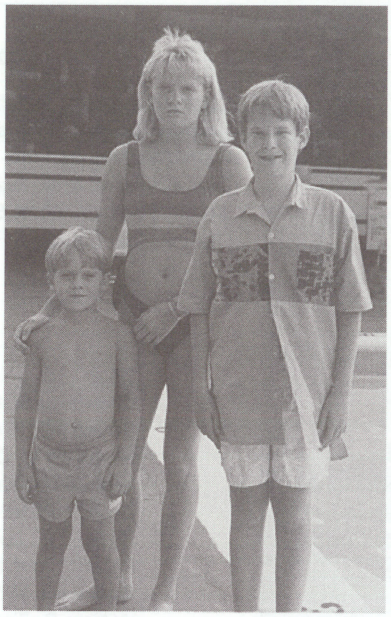
What we call a "real" vacation has very specific criteria, the first of which is: no kids. Sun, water and no mental or physical exertion are also high priorities.

Thus it must have been a moment of mutual temporary insanity when we agreed we would take three kids to the 1988 PWSA National Conference, stay in one motel room and share one bathroom for five days.

The first monumental feat was just pulling out of the driveway. This in itself took hours of hard labor, and strategically packing the car so there was enough room left for five bodies and oxygen. As the last house check was being made, we looked around at: 6-year-old Mikey (our grandson) leaping excitedly around the room; 14-year-old Sarah (with her headphones glued to her ears) sulking on the steps; and 15-year-old Matt checking the house for the 15th time for his rock collection book, repeating in an uptight voice, "I can't find…"

Al turned to me and said, "Do we really know what we are doing? Is it too late to change our mind?" I just gave a sigh of resignation and responded to the first question, "Probably not." And, "I am afraid so" to the second question.

Fortunately in this era of mini-vans and reclining seats, all three slept halfway there. During the second half of the trip, Matt decided to prove to us that he should be able to get a driver's license. He did this by reading and explaining all the road signs for the rest of the trip.



*Our kids were a lot younger when Al and I drove them to the 1988 National Conference. Pictured then are, L-R: Mikey,age 6, Sarah,14 and Matt, 15.*

About 15 miles out of Louisville, Matt declared to us that he would make friends at the conference — but that he wasn't going to have a "Prader-Willi girlfriend" because, "they can't have babies, you know."

Although 2 years ago one of Matt's laments was that he couldn't have babies, he has managed to block this out. He now declares that "when I grow up I going to date a older woman (16-19) because she can have babies. She get my food and do all the cooking." When Al told him he was too young and needed to finish school, Matt said, "I have it all figured out. I go to school in the morning and work in afternoon, I good at art you know."

Fortunately, Matt quickly forgot his vow regarding girlfriends with PWS immediately after our arrival when slim, redhaired Joyce Abell put her arm around him and gave him a book on computers. Then Ricky Lacy came up, introduced himself, and shook Matt's hand.

***Road Trip continued on page 11***

*inside*

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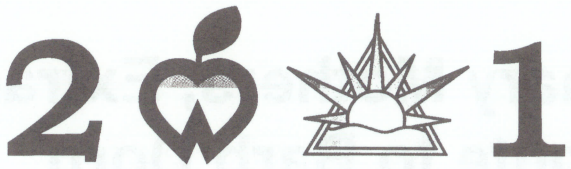
*The Gathered View* welcomes articles, letters, personal stories and photographs and news of interest to those concerned with Prader-Willi syndrome.

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.

## Japanese congressman promotes PWSA awareness

Volunteer staff of Congressman Yoshiaki Harada wear PWSA aprons as they work on his campaign in Fukuoka, Japan for re-election to the Japanese Parliament. Harada was once an exchange student in Tulsa, Oklahoma, and has stayed in close contact with his American family, Curt & Marion Shacklett and son Jon, now 32. Harada obtained 100 of the PWSA aprons for his campaign staff after a recent visit with the Shackletts in Tulsa.

# *A World Class Event*: The 2001 PWS International Conference



## June 27-July 1, 2001

### Registration

Please register ASAP to ensure that there is no confusion when you arrive. If you do not receive confirmation of registration within 2 weeks, please contact the registrar. Conference updates and registration forms are available at: *www.IPWSO.org* Registration, confirmation and registration information: *Registrar@pwsausa.org* or telephone 866-797-2872 or facsimile 941-312-0201.
Other information: *pws.world.conf@worldnet.att.net* or telephone 888-316-9869.

### YAAP Update

Science Museum of Minnesota day includes both Omni movie "Caves" and Laser Show, as well as award-winning interactive exhibits.

On Friday, June 29 there will be a Sib Shop program at nearby Saturn School for siblings who are 6 and older.

There will be a special banquet for 6- to 12-year-olds in addition to toddler and older YAAP parties.

Please remember, due to program complexities and contract commitments, no YAAP registrants are accepted after June 1.

### Scientific Conference

University of Florida will award 17 CME credits to professional attendees at no additional charge.

High score received from NIH; level of funding still pending at press time. Abstracts will be published in *American Journal of Medical Genetics.*

Scientists Dinner on Wednesday will be at Science Museum overlooking Mississippi River.

### Provider Conference

Comprehensive program includes presenters coming from six countries. PWSA of MN contributed scholarships for 15 case managers who serve clients with PWS to attend from 15 different counties.

### IPWSO Delegates, Chapter Presidents and USA Board Members

Wednesday, June 27 dinner at the Downtown University Club; invitations will be sent.

### General Conference

Thirty-five presenters, including many world-renowned scientists, come from 14 countries and 10 states.

Children will be celebrated in a special media presentation.

### Social Events

Bring your red, white and blue outfits. Friday dinner will be a "traditional" indoor 4th of July picnic. Special performance by Giorgio Fornasier and Monika Fuhrman of IPWSO at the Saturday, June 30 gala.

Note that the hotel does not have a Kosher kitchen: Kosher meals can be ordered for YAAP participants. Other meals will have a vegetarian or fish option.

### Additional Hotel Rooms

The Radisson Riverfront is filling up fast. A second additional hotel has been added at a comparable conference rate. Bookings made after June 4, 2001 will revert to standard rates.

### Recreation and Excursions

The Conference hotel overlooks Mississippi River with wonderful walking and jogging paths.
Self-planned activities may include:
Within walking distance: paddle boat trips, Children's Museum, Science Museum, Ordway Theater, Landmark Center and Saturday Farmers' Market.
Short drive: Minnesota History Center, Dodge Nature Center for children, restored Fort Snelling, Como Zoo and Conservatory, Gibbs Farm Museum, State Capital, Mall of America and historic Stillwater.
For side trips and longer excursions, see *www.IPWSO.org* for sources of information.

### Transportation Concerns

There is a shuttle service to the Conference hotel from the airport. Follow signs to transportation center. Select shuttle for Saint Paul, Radisson Riverfront Hotel. The cost of the 20-minute trip will be $16 round trip or $11 one-way. Children under age 11 are free; children under age 3 must have a car seat.

### Additional YAAP Notes

All medications to be administered during YAAP must be labeled with date, time and dosage and must be delivered to the nurse in their original containers during registration.

Remember to bring your cell phone or pager and to provide these numbers during registration.

If needed, bring a labeled bag with a change of clothes and give directly to the assistant director of the group.

Please do not bring personal belongings into the YAAP. No purses, waist or backpacks, etc. -- ***Nancy Bowman, Registrar***

***Executive Director's View***

# Ordinary Mothers, Extraordinary Lives
# A Tribute to Barb Dorn

***By Janalee Heinemann***

It is hard for me to believe that Barb Dorn has completed her 3 years as our national president. I remember when I was PWSA (USA) volunteer national president and said I was stepping down after 3 years and no one believed me. Now I find myself in the role of non-believer, but Barb has forewarned us all along that this would only be a one-term commitment for her.

As mother of a young teen with PWS, executive director of her state chapter, and a part-time teacher, Barb has devoted far more to our national efforts than we could have ever asked. She has given up many, many nights and weekends to PWSA (USA) efforts, and helped bring us to a new level of organization and chapter support.

As a member of the Executive Committee, Barb has been a great support to our board chair Ken Smith and to me. We have felt like a real team – not always agreeing, but always respecting each other's opinion.

In reflecting on Barb, a question asked of me by a volunteer last week came to mind. "Why do you do all that you do?" she asked. I look not only at myself, but Barb, Lota Mitchell (who will be taking over Barb's role, and has already worn many hats within our organization) and to many other mothers in PWSA (USA), and realize that none of us set out to do extraordinary feats.

Most of us had just planned to be ordinary mothers and wives. Our goal had not been to blaze trails and consume ourselves with a cause. But the cause came to us, and we had a choice to make. We could be beaten down by the syndrome and society's apathy – or we could learn, acquire strength and burn the midnight oil with our passion. We became comfortable with medical terms, lost our shyness to became public speakers, and found we could be smarter and work harder that we ever thought we could.

So are we unusual? No, we are just ordinary Prader-Willi Moms. We did not mean to climb the mountain, but when the mountain came between our children and us, we put on our hiking boots and began scaling it one step at a time.

I want to thank you, Barb Dorn, from the bottom of my heart. I know you need a break – like Lota and I did at one point in our lives. But I also know that like Lota and me and many others, you are just resting on a ridge for a while. Our bond and love for each other and our children will continue to push us up the mountain and beyond the ordinary.

# *Our Parent Mentoring Program at Work*

*By Carolyn Loker*

Following a referral from the PWSA (USA) office, I asked Volunteer Parent Mentor Vicki Knopf (who is also president of the Connecticut State Chapter), to contact the mother of a baby recently diagnosed with PWS. The mom has three other children under age 6. Here is Vicki's report and my response:

Dear Carolyn,

Just to let you know, I just got off the phone with the new mom. We spoke for about a half hour. Baby came home from the hospital yesterday. I am going to follow up with her on Wednesday to see how things are going. We only live about 30 miles from each other, and she has already asked about meeting us. I will keep you posted as things progress. Thanks for passing this family on to me, I will take good care of them.

I am just glad to be a part of this mentoring program. I can only imagine the relief I would have felt to have had another parent to talk to when David was a newborn. We were so all alone in it for so long.

I could feel the pain in this mom's voice today and it reminded me of just how painful those first months are. As exhausting as these phone calls can be, it is also one of the most rewarding things I do, just to be able to give back a little is all that I want. I want to make sure that no parent is ever handed a brochure for a residential placement facility at the diagnosis of their new baby as Dave and I were.

These new parents are so lucky to have this mentoring program, and we are so lucky to be strong enough to help them.

More soon, Vicki

Dear Vicki,

You are one quick lady. I just called you an hour ago and you have already called this new mom and e-mailed me back.Thank you so much for being a part of the Parent Mentoring Program. This mom is so lucky to have you. Could you have imagined being able to talk to a mom with a younger child after David was diagnosed? It would have helped me not grieve so deeply and probably would have kept me from getting clinically depressed.

Isn't this all so bittersweet? I only wish all this was in place when our kids had gotten their diagnoses. Now it's nothing to hear another parent say "My child is on GH and Coq10" and not blink an eye. The struggles that we all had to go through to get all this started, and now it's almost standard treatment.

I'm happy for the new parent and so glad parents like you can make a difference. And I will always be eternally grateful for the parents before us.

Thanks, Carolyn

***President's Message***

# Slam Dunk Memories of Our New Basketball Star

*By Barb Dorn*

I want to extend my sincere appreciation to all of you for all of your support over the past 3 years. Thanks for those notes and phone calls of encouragement. As parents of children with PWS, we share a bond like no others share. You have all become extended family. Thanks again for allowing me to be your representative in sharing your voices and wishes. We all want what is best for all who have Prader-Willi syndrome.

Over the past three years, I have been fortunate in being able to share some of the memorable moments of my family life with you. I have to admit that those articles have been the most fun to write. I think that as parents of children with PWS, we learn to treasure those special times. My son Tony has taught me to appreciate so many things in life.

*Basketball player Tony Dorn*

This is my last message to you as your president. Once again I want to share a special time that our family recently experienced. We have been experiencing a lot of "firsts" this year. One that has been especially fun has been Tony's involvement in the Special Olympics program. This winter, he played on the local basketball team. We were all very proud,but Tony was the most proud.

Our family loves sports. We enjoy watching and participating in almost any of them. But Tony never seemed to find one that he enjoyed playing and could personally succeed at. I recall when he was younger he tried T-ball. We found however that we had to spend a great deal of time getting him to remain standing and not to sit in the outfield. He loves to swim, but he wasn't skilled enough or fast enough to compete. Most Special Olympic events were scheduled later in the evening, a time that isn't typically Tony's best time. Tyler, Tony's brother, has been a sports participant since he was very young. Tony has been the loyal cheerleader (at least when his mood was good) for many years. It has been a pleasant change to have Tony out on the court and see his brother in that cheerleading role.

Tony was the youngest member of the team. He had never played competitively before. It was going to be a year of learning. His teammates weren't afraid to get in there and be aggressive. Tony needed some work in that area, but he did his best. We were all truly amazed to watch him race back and forth down that court. I know I could never get him to exercise like that. His effort as well as the effort of all his teammates was remarkable.

One of the home games was scheduled in the evening, and they played it in their high school gym, so friends and neighbors attended. Thanks to the efforts of his coach, you had to check closely to see what team was playing – the varsity team or the Special Olympics team. Some of the varsity players served as referees. The pep band played and the cheerleaders cheered. The stands were about half full of fellow students from their high school. It had the atmosphere and enthusiasm of a final game in a big playoff series, and it brought tears to our eyes. We were so proud.

Tony's team did not have a winning season. It didn't matter. Each game was a win in its own special way. We all want our children to be happy, have fun, be accepted and succeed. This Special Olympics team was our first experience with this program. All of those wishes came true. As a family, we look forward to many more of them in the future. Take care.

**As parents of children with PWS, we share a bond like no others share. You have all become extended family. Thanks again for allowing me to be your representative in sharing your voices and wishes.**

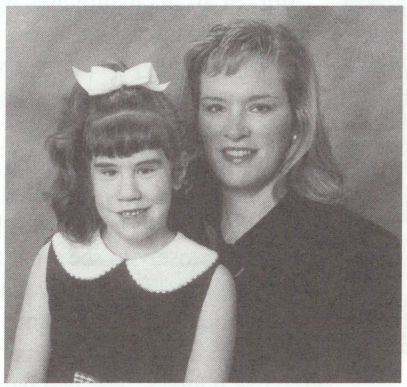
# Candidates for the PWSA (USA)

## About the Candidates

*Proxy Forms are printed on pages 13-14*

The Leadership Development Committee has put forth the following slate of candidates for consideration by the membership. There are five nominees for four seats on the Board. Voting for candidates takes place in person at the National Conference in June 2001 or by proxy. All nominees were invited to submit a statement and photograph. Their submissions follow, in alphabetical order.

### *Kellie Aragon, Dallas, Texas*



*Kellie Aragon and daughter Natalie*

I am a part-time certified public accountant. My husband Michael and I have three wonderful children. Our middle child, Natalie, now 5 years old, was diagnosed with PWS at age 2.

We quickly became involved with the Texas PWSA Chapter and joined the PWSA (USA). I am the current vice president of the Texas Chapter and Michael is on the Board of Directors.

With the Texas Chapter I have helped to establish PWS awareness and parent support. To help facilitate awareness locally, I worked with *The Dallas Morning News* to do an article about PWS that had great results. I am currently working with the Texas board for legislative change to aid our children. I am also a council delegate for PTA and am active in church and local schools.

Our family has attended two conference since our PWS diagnosis, and we plan to be in Minnesota. From these conferences I have gained knowledge and respect for the PWSA organization.

I would like the opportunity to serve you, the membership, as a member of the PWSA (USA) Board of Directors. If elected to the Board, I will be an advocate for you and your children, as well as offer my business experience to the organization. Together, we can ensure that our children with PWS have the best opportunity to live life to the fullest.

### *Daniel J. Driscoll, M.D., Ph.D., Gainesville, Florida*

*Dr. Dan Driscoll and a patient*

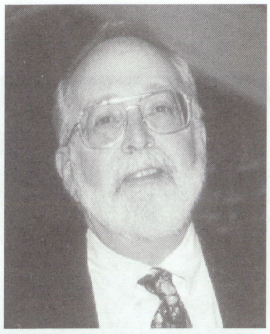
Dr. Driscoll is a professor of pediatrics and molecular genetics at the University of Florida College of Medicine. He completed medical school at Albany Medical College and received his human cytogenetics training while pursing his doctoral dissertation at Indiana University. His pediatric residency and clinical genetics fellowship were done at the Johns Hopkins Hospital, where he also received his training in molecular biology, particularly DNA methylation.

He has had a long-standing interest in the Prader-Willi syndrome and the phenomenon of genomic imprinting. His laboratory has made several important contributions to the field, including being the first to propose DNA methylation to diagnose the Angelman and Prader-Willi syndromes. His clinic closely follows more than 100 individuals with Prader-Willi from all parts of Florida and southern Georgia.

Dr. Driscoll is board certified in pediatrics, clinical genetics, molecular genetics and cytogenetics.

He has served PWSA (USA) in a number of capacities, including as a current member of the Board of Directors, co-chair of the Clinical Advisory Board and co-chair of the Scientific Conferences at the 2001 PWS World Conference.

# Board of Directors Election



*Jim Gardner*

### *James P. Gardner, White Bear Lake, Minnesota*

Jim has been a member of the Board of Directors of PWSA (USA) and the association's treasurer since 1995. He and his wife Joan are co-chairs of the 2001 PWS/IPWSO Conference in Minnesota. At the chapter level, he has served as president of the Prader-Willi Association of Minnesota, Inc., since 1992.

He is the father of Lawrence, 26, who has Prader-Willi syndrome.

Jim is currently chairman of the board of Little Mountain Bancshares, Inc., director and loan committee chair of First National Bank of Monticello/Lakeville and proprietor of Gardner Management Co.

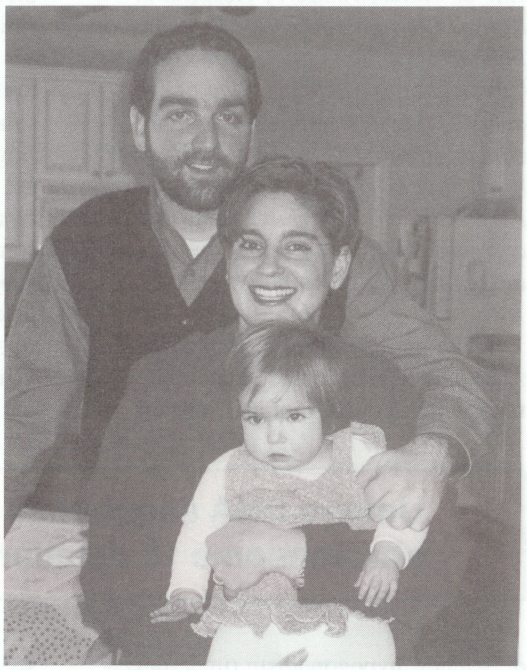
In addition to his work for PWSA, Jim has held offices in United Cerebral Palsy of Minnesota, Inc., and of Greater St. Paul, as well as a number of other civic and charitable organizations.

### *Robert Lutz, Cincinnati, Ohio*

My only child — Isabel — was diagnosed with Prader-Willi syndrome (uniparental disomy) approximately 1 year ago, when she was 5 months old. My family benefited greatly from the PWSA during the tough time when we first learned of Isabel's condition. Attending last year's conference, reading PWSA materials and connecting with other PWS parents made a huge difference in my family's ability to understand the challenges we face and to give Isabel the best start possible. Through that process, I came to deeply appreciate and respect the mission of the PWSA and I would like to contribute to that mission by volunteering my time, energy and experience.

I believe that my business education and experience will enable me to make valuable contributions both raising money to fund the PWSA and PWS research as well as making the results of that research meaningful to all PWS families.

I am currently chief financial officer of a division of Cinergy Corp., an $8 billion electric and gas utility company. I have an MBA from the Kellogg Graduate School of Management, Northwestern University, and business board experience as a non-voting board member of Q-Comm, a $100 million telephone company in Indiana. I have also served as a volunteer in the non-profit arena.



*Bob, Debra, and Isabel Lutz*

*Ken Smith*

### *Ken Smith, Pittsburgh, Pennsylvania*

Ken has worked with people who have Prader -Willi syndrome and other disabilities for 16 years at the Children's Institute (formerly the Rehabilitation Institute) in Pittsburgh, Pennsylvania.

He began at the Institute working in direct care with people who have PWS. Then for 9 years he was an assistant to Bea Maier, one of the program's founders, who retired in 1994. Ken has served as the program's manager for the past 6 years.

Ken has served on the PWSA (USA) board of directors for 5 years and is completing his second term as Board chair.

If re-elected, Ken looks forward to continuing efforts to help PWSA (USA) better serve individuals with PWS and their caregivers.

# Correlating Genetic Research with Clinical Findings

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

*In research lies our hopes for successful management and treatment of our children and adults with Prader-Willi syndrome. In the first of a two-part article, Merlin G. Butler, M.D., Ph.D., FACMG, who is Chairman of the PWSA(USA) Scientific Advisory Board, provides an overview of some current research and research findings.*

*Dr. Butler is chief of the Section of Medical Genetics and Molecular Medicine at Children's Mercy Hospital in Kansas City, Missouri, and Professor of Pediatrics at the U. of Missouri-Kansas City School of Medicine. One of only about 500 cytogeneticists and 1,000 clinical geneticists in the U.S., he co-headed an extensive research project on PWS while at Vanderbilt University. He is currently seeking major funding from NIH to continue the research efforts.-Ed.*

The main features of Prader-Willi syndrome (PWS) include hypotonia (weak muscle tone) during infancy, mental deficiency, hypogonadism (underdeveloped sex organs), behavior problems, early onset of childhood obesity, small hands and feet and a characteristic facial appearance.

Approximately 70 percent of individuals have a deletion of the proximal long arm of chromosome 15 (called the 15q11-q13 region) donated by the father. About 25 percent have maternal disomy of chromosome 15 (instead of one chromosome 15 from the father and one chromosome 15 from the mother, both chromosome 15s are from the mother). The remainder have biparental (normal) inheritance of chromosomes with mutations of the imprinting controlling center which regulates the activity of genes in the 15q11-q13 region, or other genetic abnormalities involving chromosome 15 (e.g., chromosome translocations).

The incidence of this condition is 1 in 10,000 to 25,000 individuals. It is estimated that there are 350,000-400,000 people with this syndrome worldwide.

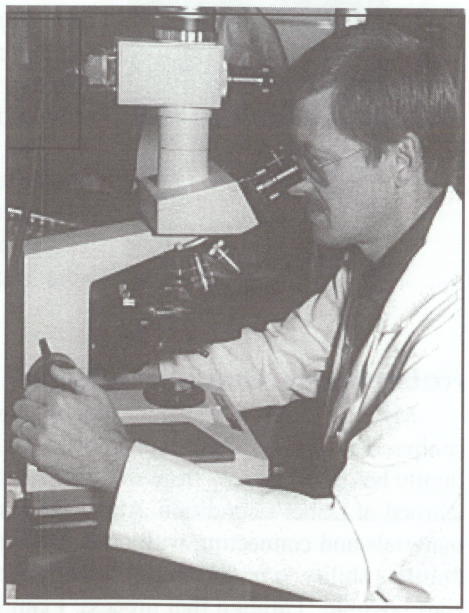
**Genetic Classes of PWS**

There appear to be five main genetic classes of Prader-Willi syndrome. First, approximately 70 percent have a paternally derived deletion from the 15q11-q13 region; second, 25 percent have maternal disomy 15 (both intact chromosome 15s from the mother).

A third class, termed imprinting mutations (less than 5 percent), have very small deletions in the imprinting controlling center of the 15q11-q13 region, or are sporadic in nature with no deletion of biparentally (normal) inherited chromosome 15s. A subset of these unusual subjects with a submicroscopic or atypical deletion can be detected with fluorescence in situ hybridization (FISH) analysis using classical 15q11-q13 DNA probes, but other subjects require additional specialized testing with chromosome breakpoint analysis and DNA sequencing studies.

A fourth class of subjects with features of PWS are those with a balanced reciprocal chromosome translocation, involving the 15q11-q13 region and distinguished from the unbalanced translocations that may lead to maternal disomy 15 or produce 15q11-q13 (or large) deletions. This rare class probably accounts for less than 0.3% of PWS subjects.

The fifth class may be those with structural gene mutations of the 15q11-q13 region. This class is theoretical and has yet to be identified in subjects. Targeting these unusual PWS subjects (those with imprinting mutations, reciprocal translocations, or possibly structural gene mutations of chromosome 15) and studying expressed genes from the 15q11-q13 region may allow for a better understanding of the effect of genetic anomalies on the clinical presentation and the location of candidate genes for specific clinical features. This is an important area of research under way at several large genetics centers.



*Dr. Merlin G. Butler*

**Unique DNA sequences may play a role in producing the common chromosome 15 deletion seen in both PWS and Angelman syndrome.**

**Prader-Willi Syndrome and Angelman Syndrome**

Prader-Willi syndrome and Angelman syndrome, although entirely different, are unique clinical disorders that most often arise from a deletion of 3 to 4 million DNA base pairs comprising genes from the 15q11-q13 region during sperm production, causing PWS, or during egg production, causing Angelman syndrome. PWS and Angelman syndrome were the first examples reported in humans showing that genetic information is expressed differently depending on the parent of origin. This phenomenon is

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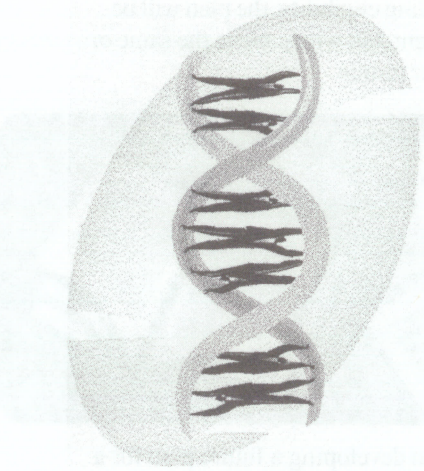
**There is a battery of genetic testing available now to determine genetic status...**

called genetic imprinting.

Recent studies have shown unique DNA sequences that are repeated and located at specific areas of the 15q11-q13 region. These DNA sequences may play a role in producing the common chromosome 15 deletion seen in both PWS and Angelman syndrome. Environmental or other genetic factors may affect genetic information in sperm or egg production, leading to the chromosome 15 deletion seen in the majority of patients with these two syndromes. Further genetic studies are under way to identify these factors.



**Recurrence Risk**

Although the recurrence risk is generally low, the molecular genetic cause may impact significantly on recurrence risks. For example, in a rare family the father may carry an imprinting mutation or a very small deletion of the imprinting controlling center on his chromosome 15 (received from his mother), but he is unaffected. The genes causing PWS on chromosome 15 from his mother are generally silent (imprinted). He then could pass on the chromosome 15 from his mother with the mutation to his child and PWS would result. Genetic information must be intact and normal from the father's chromosome 15 to avoid not having PWS in the child. This could occur at a 50 percent risk.

These family members will require more detailed laboratory genetic testing than available in the routine cytogenetic or molecular genetic laboratories. There is a battery of genetic testing available now to determine the genetic status (large deletion, small deletion or imprinting mutation, maternal disomy, or chromosome translocation), and services of a genetics center would be recommended to assist in establishing the exact genetic cause and to supply genetic counseling and healthcare management.

**Research on Genes Related to PWS**

There are at least one dozen genes (segments of the DNA molecule) identified in the 15q11-q13 region. However, this region may contain about 100 genes; therefore, more research is needed to identify other genes and to determine the role of these genes, when deleted, that cause PWS. The genes causing PWS must be able to be turned off or on depending on the parent from which the genes were received. The genes causing PWS are generally active (expressed) only on the chromosome 15 from the father and inactive on the chromosome 15 from the mother. These paternal genes are deleted or abnormal in the PWS individual.

So far, five imprinted or paternally expressed genes (ZNF127, NDN, MAGEL2, SNURF/SNRPN and IPW) have been localized within the 15q11-q13 region.

Additional genes that are apparently active on both the mother's and father's chromosome 15 are also found in the 15q11-q13 region, including receptor genes for gamma butyric acid (GABRB3, GABRA5 and GABRG3), an important neurotransmitter protein found in the brain and the P gene involved in skin pigment production. Since the 15q11-q13 region is relatively large and visible under the microscope, it is likely to contain more than one paternally expressed gene involved in causing PWS.

**Identification of additional genes in the chromosome region and their role in causing PWS is an active area of research which may be quite important, specifically for pharmaceutical approaches or replacement of proteins needed for treatment of those with PWS.**

**Further Research on Specific Genes**

The best characterized paternally expressed gene on chromosome 15q11-q13 region is SNRPN (Small Nuclear RibonucleoProtein N). A second region of this gene is termed SNURF (SNRPN Upstream Reading Frame), which is considered to be the imprinting center. This genetic locus appears to have a key role in the regulation of imprinting (controlling activity of other genes) throughout the 15q11-q13 region. Disruption of this locus will cause the loss of function of paternally expressed genes in this region.

Furthermore, evidence from those rare patients with typical features of PWS with chromosome 15 translocations (rearrangements) involving the 15q11-q13 region and particularly of the father's chromosome 15 shows disruption of the SNURF-SNRPN gene. Those with translocations involving the 15q11-q13 region but not disrupting the SNURF-SNRPN gene may have some features of PWS but are not typical. Studying these rare individuals (16 have been identified since 1990) with translocations of the 15q11-q13 region should give us useful clues in identifying specific DNA sequences or genes that code for proteins, which, when disrupted, cause features of PWS.

*Research continued on page 10*

# The Family Trust: A Resource for Individuals With Disabilities and Their Families

*By Timothy Pawol*

This is the second of a two-part series on planning considerations for individuals with disabilities and their families.

Families of individuals with disabilities face a number of challenges in planning for the future of their disabled loved ones. Families will ask such questions as Who will oversee the well-being of my family member when I am gone? and How do I provide for future financial support for my family member without jeopardizing his eligibility for public benefits?

The Family Trust, a non-profit corporation sponsored by the ARC Allegheny system, has set a goal that families should have the same option to design and fund a future plan for a family member with disabilities as for any other family member. For various reasons, including eligibility, the plan will be different, but will achieve the same or similar results.

In developing a future plan for a disabled family member, families need to consider what supports will be available from the government, what supports the family will fund to supplement the governmental services and the amount of funds necessary to continue the supplemental supports for the future. In addition, the family should consider whether assistance with decision making is necessary or desirable. If the family finds that assistance is necessary, it should examine the options of representative payee, power of attorney, advance medical directives, executor, trustee and guardian. Each option has its advantages, disadvantages and limitations and should be considered in the context of the goal to be achieved.

The Family Trust is available for any person with a disability throughout the Commonwealth of Pennsylvania and is willing to assist families or organizations in other areas to establish their own trusts. The Family Trust may be reached at 412-995-5000 ext. 405 or 434, or at 711 Bingham St. Pittsburgh, PA 15203 or via e-mail at *www.arcallegheny.org.*

## Research - *continued from page 9*

**Animal Model Research**

The use of animal models has been helpful in studying human disorders. For example, in mice the genetic information from the central region of mouse chromosome 7 is similar to the proximal long arm of human chromosome 15. Genetic mouse models with comparable chromosome deletions found in humans with PWS, such as excess maternal contributions or single gene deletions, offer the opportunity to investigate how genetic defects affect behavior, brain neurochemistry and anatomy in animals; this would establish parallels with PWS in humans. Currently, single specific genes missing in subjects with PWS have been localized and removed from mouse chromosomes, and these mice do not show the features of PWS. However, when several genes equivalent to the 15q11-q13 region in humans are deleted in mice, the mice are born weak, hypotonic and die usually within the first week of life. Additional studies are under way to further design and study animal models for PWS investigations.

**Total energy expenditure (amount of calories used by the body), physical activity and amount of muscle mass appear lower in PWS subjects compared with obese individuals.**

**Research on Obesity**

Obesity, the syndrome's most significant health problem, is due to hyperphagia (overeating), physical inactivity, decreased metabolic rate and inability to vomit. Marked weight gain and life threatening obesity may occur and have been the focus of several research investigations.

The unusual fat patterning (males with PWS have more subcutaneous fat than females), which is centrally located in PWS individuals, may make certain health problems such as diabetes more likely. Studies have shown that the amount of subcutaneous fat (under the skin) versus visceral fat (inside the abdomen around the organs) may also impact on overall health.

There appears to be no difference in the amount of subcutaneous versus visceral fat in subjects with PWS or those with simple obesity, but visceral fat deposits may be regulated differently in PWS subjects than obese individuals; this may affect fat metabolism in PWS.

Total energy expenditure (amount of calories used by the body), physical activity and amount of muscle mass appear lower in PWS subjects compared with obese individuals. The decreased muscle mass and related decreased physical activity are probably major factors leading to the decreased energy expenditure in PWS.

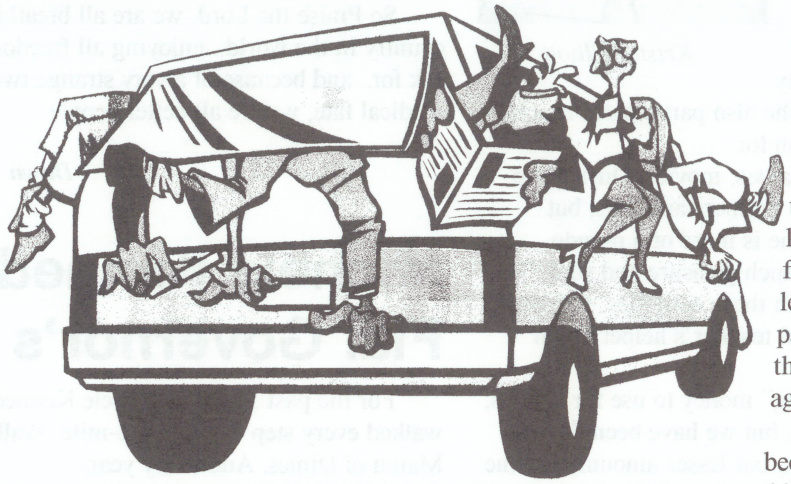
More studies are required on energy balance and factors that may improve metabolism and energy expenditure, for example, growth hormone use and time of treatment.

- continued from page 1

and shook Matt's hand.

Kentucky, being the friendly chapter we know them to be, has also bred friendly children. They all helped us unload the van and within an hour Matt had declared Joyce his girlfriend. Later he looked up at me quizzically and asked, "Is Joyce Prader-Willi, Mom?" I replied, "Well, yes she is. Isn't she pretty though?" Matt had to admit she was.

I wish we could say Matt's issues of acceptance of himself and others with the syndrome ended there, but by the next morning Matt had met some of the "normal" sibling counselors and was enamored by their beauty and what they represented to him. Throughout the rest of the conference Matt vacillated between feeling like the best of the kids with PWS to hovering in the shadows of the teenage siblings.



Matt beamed frequently when people told him how good he looked and when Lota Mitchell, board chairperson, asked him if he knew he was the most famous kid with PWS in the nation. Matt acknowledged that indeed he knew that and signed his autograph for her.

On the other hand, when Sarah was going to the sibling group he asked, "Why I can't come? What they talk about? They think I too stupid to have conversation?"

Later, at the dance, when we walked in, Matt was smiling and dancing with a slim young woman with PWS. When I went to take his picture he immediately stopped dancing and grabbed two attractive teen siblings to have his picture taken with. Matt is not entirely alone with his self-image dilemma. Curtis Deterling, another 15-year-old with PWS, also proudly showed me the addresses of the same two "normal" girls. Matt pulled me aside and confidently whispered that they liked him better. The boys' struggle can sadly be summed up with the following example. When we introduced Curtis to Sarah he asked her, "Are you Prader-Willi?" When she said no, he apologized profusely for even thinking that she might have PWS — like himself.

The conference was, unexpectedly, the most significant for Sarah. At 14 her biggest goal is to be one of the "in crowd" and there is nothing "in" about having a brother with Prader-Willi syndrome. Recently she had been showing signs of shame and intolerance with Matt.

In Kentucky though, Sarah suddenly found a new community of "blood brothers and sisters" and a new sense of acceptance of Matt. Along with other siblings her own age, Sarah told of her concerns and frustrations. The sibs all slipped each other snacks, shared funny stories and formed special relationships. Sarah also received many positive comments of recognition for the sibling book that she and I wrote half her lifetime ago. It suddenly became okay to be a part of a Prader-Willi family again.

We included our 6-year-old grandson, Mikey, in our local PWS activities and retreats and brought him to the conference with us because he, too, must adapt to the syndrome. We feel it can't hurt him to learn to be comfortable around people who may appear unusual to others. Locks in the kitchen, not being able to eat snack foods in front of Matt, and not leaving leftover food around are all a part of life for Mikey. All of these things he learned at a very young age.

Now at age 6, Mikey is becoming more aware that some things are unusual about Matt. He recently asked, "Matt, why do you always ask questions?" and, "Matt, why do you always worry about your things so much?"

One of the special positive aspects of the National Conference is that it truly is geared for all ages. Mikey, along with Sarah and Matt, danced every dance on Friday night. He found a 7-year-old girlfriend, Sayward, whose room number he promptly wrote on a piece of paper (the result of being around teenagers.)

**Matt beamed frequently when people told him how good he looked and when Lota Mitchell, board chairperson, asked him if he knew he was the most famous kid with PWS in the nation.**

Adding a fourth generation to our Kentucky experience, Al's parents from Iowa surprised us with a two-day visit at the conference. When discussing the impact of PWS, we usually focus on the immediate family, but the effect of PWS encircles a much larger group of people in varying degrees. The extended family involvement of the Kentucky Chapter in assisting with the conference was heartwarming. The people brought into the PWS circle of awareness and support grows each year.

So, on the way home, as we all basked in the warm glow of the conference, Al and I were breathing a sigh of relief that our group adventure had gone far better than our most optimistic expectations. The only hitch was when the kids all said they had so much fun that they wanted to go next time! Al and I cringed, looked at each other with pained faces and said in unison, "Three kids? All the way to Canada?"

# A Twist of Medical Fate Has Made Us Better People

My daughter Kristin will be 20 on May 15. After graduating from Kamiakin High School last year, Kristin is now in her second quarter at our community college here in Tri-Cities, Columbia Basin College. She takes two classes per quarter, math and an English-related. She gets to and fro via our Dial A Ride service.

*Kristin Elliott*

She still volunteers at The Life Care Center, a skilled nursing facility for the elderly here in Kennewick. She also participates in a lot of activities put on by the Association for Retarded Citizens, dances, hockey games, movies. During the summer she is officially a camper at Partners and Pals, but because she is so high functioning she is more of a pseudo counselor, from which she derives much pleasure and some much needed self-esteem. Along with these activities, Kristin is a volunteer Preschool Sunday School teacher's helper at our church.

Due to Kristin's habit of "finding" money to use for edibles, it is impossible to allow her to work, but we have been fairly successful in allowing more frequent, but lesser amounts of time each time in her volunteering efforts so as to cut down on the amount of food she might acquire. So far, so good.

I feel her volunteering is our way of paying back the generous support the government affords us with her SSI funds and medical coverage. (Praise the Lord we were successful in obtaining SSI for Kristin.) My husband and I make a decent living, we live in a nice home, we drive nice vehicles, but alas, we are typical Americans, we spend probably 10 percent more than we make! There never seems to be enough for everything, so that SSI has really helped alleviate some pressure with the cost of Kristin's college expenses and social activities.

She also benefits from Federal funding of our Division for Developmental Disabilities, which is based upon the severity of the handicap and not on the family income. They give a set amount per year for things that enhance the quality of life for special folks. Kristin uses it for a membership at a local health club, which is fabulous, along with paying for the Partners and Pals summertime camp program and the occasional dental things that insurance and/or SSI do not cover.

Kristin is still very into her cross stitching, her Pomeranian, Toby, and her computer. If anyone ever wants to email her she would love it... her address is: *Kristin 0581@aol.com*

Kristin has brought great joy and great sorrow to our family through the years. We have all had to adapt our lives and hearts to her needs. It is not always a Norman Rockwell family that you see in action at our house, but even through the roughest times, perhaps most of all at those times, we are bound together by the weight of the restrictions Prader Willi syndrome has brought to us.

Our love for one another is never stronger than when we are meeting one of a myriad of needs brought about by the related crises that are a constant with Prader Willi syndrome. Not a week goes by that Kristin doesn't amaze and infuriate us with some new thing she has accomplished or figured out how to manipulate!

We are on a regular schedule of changing the changeable combination locks that have been a staple in our kitchen since she was 4 years old. It gets kinda tricky if you don't remember to change all the locks at the same time!

So Praise the Lord, we are all breathing, living in the best country in the world...enjoying all freedoms we could possibly ask for...and because of a very strange twist of medical fate, we are all better people.

***Dawn Elliott, Kennewick, WA***

# Michele Kennedy Wins Fla. Governor's Award

For the past 5 years, Michele Kennedy of Seminole, Fla. has walked every step of the 8-1/2-mile Walk America event for the March of Dimes. And every year, she's raised the most money for Alachua County's Association for Retarded Citizens.

On March 15 this year, Michele won the Governor's Point of Light Award for her efforts. "Michele is an inspiration to others, not only for working so diligently to conquer her own challenges brought on by Prader-Willi syndrome, but also for her contributions to the March of Dimes Walk America," wrote Sara Struhs of the Governor's Points of Light Awards.

In October, Michele will go to Tallahassee for lunch with Florida Governor Jeb Bush to receive her award in person. In the meantime, Michele is listed as a winner on the Florida Commission Community Service web site: *www.fccs.org.*

*Michele Kennedy*

Since joining the Alachua County ARC in 1995, Michele has lost more than 150 pounds as she has accelerated her efforts each year to raise money for the March of Dimes.

Michele is the daughter of David and Jody Kennedy of Seminole, Fla. "We are so proud and happy for her," said Jody Kennedy of her daughter.

"You should be very proud of your daughter; she is a true Point of Light," Struhs told the Kennedys.

# Instructions for Completing the Proxy Ballot

Note: The official proxy ballot is on the next page. Use the proxy ballot to cast your vote if you are a PWSA (USA) member in good standing and you will not be present at the Minneapolis Conference to vote in person. Family memberships may submit two proxies.)

**Your proxy form will be hand-delivered to the person whose name you write here** (any eligible voter who is attending the conference, including any current officer or board member).

**The state name will help us identify the member you've chosen as your proxy, if it is not a current officer or board member.**

Prader-Willi Syndrome Association (USA)

## 2001 Official Proxy

I hereby appoint _____ of _____ to
(print name of your designated proxy voter) (state of residence)

vote as my proxy at the PWSA (USA) Annual Membership Meeting in Minneapolis, Minnesota, June 2001.

Instructions to proxy voter:

☐ **Please cast my vote as you see fit** **OR**

☐ Please cast my vote for the Board of Directors candidates I have marked below:

☐ Kellie Arragon

☐ Daniel J. Driscoll

☐ James P. Gardner

☐ Robert Lutz

☐ Ken Smith

☐ _____
(This space provided for a write-in candidate)

I am a member in good standing of PWSA (USA)

Name _____
(please print)

Signature _____

Date _____

**If you wish to have your designated proxy voter choose board candidates, check the first box, and don't check any others on the ballot.**

**Please print clearly on the Name line, using the name that appears in our membership records, then sign your name on the Signature line the way you sign other legal documents.**

**Cast your vote for chosen board candidates by checking this box under "Instructions to proxy voter" AND checking the box next to each of your selected candidates.**

**PROXY FORMS MUST BE RECEIVED AT THE NATIONAL OFFICE BY FRIDAY, JUNE 15, 2001**

MAIL TO: PWSA ELECTION, 5700 Midnight Pass Rd., Suite 6, Sarasota, FL 34242

## Prader-Willi Syndrome Association (USA)
# 2001 Official Proxy

I hereby appoint _____ of _____ to
(print name of your designated proxy voter) (state of residence)

vote as my proxy at the PWSA (USA) Annual Membership Meeting in Minneapolis, Minnesota, June 2001.

Instructions to proxy voter:

☐ **Please cast my vote as you see fit** **OR** ☐ Please cast my vote for the Board of Directors candidates I have marked below:

☐ Kellie Arragon

☐ Daniel J. Driscoll

I am a member in good standing of PWSA (USA)

☐ James P. Gardner

Name_____
(please print)

☐ Robert Lutz

Signature_____

☐ Ken Smith

Date_____

☐ _____
(This space provided for a write-in candidate)

---

## Prader-Willi Syndrome Association (USA)
# 2001 Official Proxy for Second Family Member

This ballot to be used by Family Memberships only

I hereby appoint _____ of _____ to
(print name of your designated proxy voter) (state of residence)

vote as my proxy at the PWSA (USA) Annual Membership Meeting in Minneapolis, Minnesota, June 2001.

Instructions to proxy voter:

☐ **Please cast my vote as you see fit** **OR** ☐ Please cast my vote for the Board of Directors candidates I have marked below:

☐ Kellie Arragon

☐ Daniel J. Driscoll

I am a member in good standing of PWSA (USA)

☐ James P. Gardner

Name_____
(please print)

☐ Robert Lutz

Signature_____

☐ Ken Smith

Date_____

☐ _____
(This space provided for a write-in candidate)

# Individuals sought for NIH liver study

Pediatric gastroenterologist Ann O. Scheimann is currently developing a program project grant proposal for the National Institutes of Health (NIH) to study steatohepatitis (fatty infiltration of the liver).

Dr. Scheimann treats both children and adults with Prader-Willi syndrome in the Baltimore/Washington area and as adjunct faculty at Texas Children's Hospital in Houston, Texas.

Within the PWS clinic population at Texas Children's Hospital, Dr. Scheimann has found abnormal liver enzymes and abnormal liver imaging (ultrasound, etc.) in a number of children with PWS.

To develop her study, Dr. Scheimann is asking that individuals with PWS who have had the following contact her as soon as possible:

1. Liver enzymes testing (normal or abnormal -AST, ALT, Alk Phos, GGT, Bilirubin)
2. Abnormal imaging (ultrasound, CT scan, MRI)
3. Liver biopsy
4. Urine organic acids or plasma carnitine studies

If there is a significant number of individuals with PWS with abnormal studies, Dr. Scheimann is very interested in designing an NIH-funded diagnostic/therapeutic study as part of the grant application.

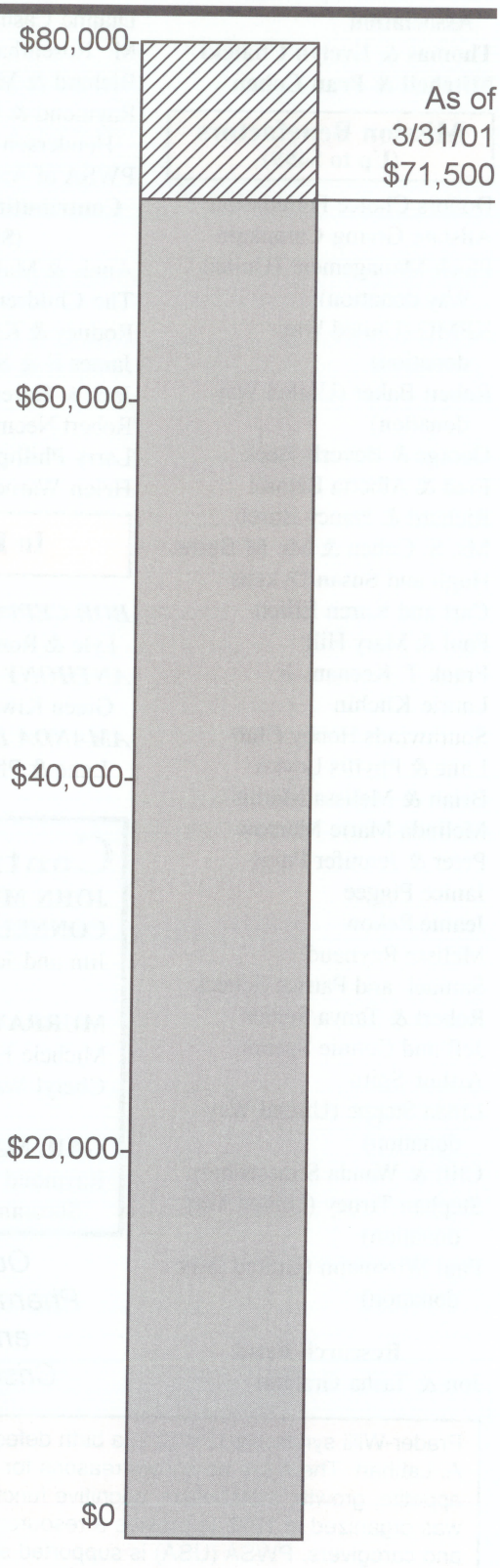
Her deadline for submission to the NIH is July 14, 2001.

Contact her as follows:

Ann O. Scheimann, M.D.
600 N. Wolfe St.
Johns Hopkins Children's Center
Brady 320
Baltimore, MD 21287
Telephone: 410-955-8765
Pager: 410-283-4900

**Angel Fund 2001**

**OUR GOAL**
**$80,000**




## The Chuckle Corner

One of the adult men with PWS who lives in our residential program in northeast Pennsylvania flies home to West Virginia a couple times a year. He's very capable of handling the travel, which includes transferring planes in Washington, D.C.

Still, when he didn't arrive as anticipated after a visit, we were worried. Worse, the airline would give us no information as to what flight he would be arriving on, as we are only "staff," not family.

Finally he called and we learned that, either through tight scheduling by the airline, or simply because he moved too slowly between his arrival and his departure gates, he had missed his connection in D.C. and would be taking a later flight back.

His roommate overheard us anxiously discussing the challenges of being alone in a strange airport. Also from West Virginia, David quickly reassured us with a knowing grin: "Don't worry; when you miss your flight, the airport has to give you food coupons."

***Joy Smith, Keystone Community Resources, Inc., Scranton, Pa.***

***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.***

# Acknowledgements

***Our Sincere Thanks for Contributions Received Through March***

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*Our thanks for ongoing support from Pharmacia for our Triage Support Specialist and the Alterman Foundation for our Crisis Intervention/Prevention Counselor*

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