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Gathered View

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



Focus on Fund Raising:

New Sources of Funds Will Help PWSA (USA) Meet Growing Needs

by James G. Kane, Chair, PWSA (USA) Board of Directors

One of the most difficult challenges for any association, company, or even household in these turbulent times is ensuring that there is enough money to make ends meet and also to be able to take on new projects considered to be truly worthwhile. Throughout the past several years PWSA (USA) has operated at a small deficit, while falling somewhat behind in delivering the products and services that it set out to provide to its members and to the professional community at large.

The fast pace of change in today's world, particularly within the medical field, and the demand for timely information from our members has forced us to accelerate our efforts to serve the PWS population. Therefore, I would like to announce two new programs to raise substantial capital to fund the operations of PWSA (USA) and many of the special projects that we all believe are critically important.

What is unique and exciting about these two programs is that the targets for fund raising are outside the membership and our traditional sources. The board and officers are sensitive to protecting our members from an unending stream of requests for funding to support the Association. We will continue to conduct our annual fund-raising campaign in the fall, the very successful T-shirt and greeting card sales efforts, our membership drive, and the sale of educa-

tional materials. Those efforts involve our membership in the Association's progress and show the outside world our true depth of interest.

Liaison Program to Build New Partnerships

The Liaison Program is a coordinated effort of the PWSA (USA) board of directors, its officers, and members to establish long-term relationships with foundations, corporations, and interested parties across the country. Through the program we will spread the understanding of PWS into a community which heretofore was unaware of the syndrome and its implications. Through that understanding we will build an extensive network of relationships.

The spectrum of organizations for contact includes pharmaceutical companies either doing research on PWS or providing drugs or equipment for the treatment or management of the syndrome, foundations or corporations that provide philanthropic assistance, and companies producing entertainment products such as puzzles or crafts particularly exciting to our children and adults with PWS.

The key to the Liaison Program is the combination of personal involvement with that outside organization by an individual PWSA (USA) member and a national board

member or officer. This approach will result in personal, direct contact and long-term continuity—key elements in successful education and fund raising. The board has already begun this program through its own personal contacts and those of several other PWSA members.

If any member has a contact in an organization that we should consider approaching, please give me a call at 410-321-9788 (office) or 410-494-8079 (home). Working with Janalee and Russ, the board will pair that member with an appropriate board member or officer to plan the best approach to the new group.

Grant Committee Formed

In today's world of extensive due diligence and complicated paperwork, the process of requesting philanthropic grants has gotten very complicated. In most instances a formal written grant request is required. Grant requests may consist of three or four pages of grant specific material plus 20 to 25 pages on the organization, its history, and its financial position. We have organized a standing committee, the Grant Committee, to produce these requests in a streamlined manner.

We have already received grants through the efforts of the Atlanta Conference Team and have submitted a request for funds to The Ronald McDonald Children's Charities. *What we need is help from our members in locating new sources of grants and help on the new committee to produce formal grant requests.*

Volunteerism is the backbone of PWSA (USA)! Please help your board and officers help you!! Contact the National Office for more information or to volunteer:

1-800-926-4797.

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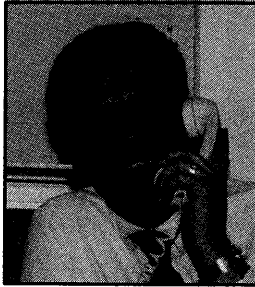
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Out of The Office

by Russ Myler, *Executive Director*

One major goal of PWSA (USA) is to educate people about Prader-Willi syndrome. The results of all our efforts are certainly paying off. Incoming calls to the National Office for the first three months of 1994 have almost doubled the first three months of last year (953 compared with 527). Most of the calls are requests for information. The mailed requests have tripled! While the volume of requests has put quite a strain on our ability to respond as promptly as we wish, we are excited about the increase.

Another goal is to extend service to families. New members joining the Association are averaging over 20 a month. Of the former members contacted since the first of the year, 7 out of 10 have rejoined the Association. The PWSA (USA) family is growing.

Moving us further along toward our goals is the organization of the New Jersey Chapter. We are pleased with their affiliation and plan a recognition ceremony at the annual meeting. They may be joined by Louisiana and Maryland since there are groups in formation that might be ready to join us by July.

Chapter activity is increasing. Conferences, workshops, and education programs are becoming yearly events. Referrals of persons to chapters by the National Office is a daily event. These referrals not only include new members but also professionals, government officials, and media staff. In other examples of chapter activity, the Connecticut Chapter is currently engaged in a major legislative effort, and I spent a considerable amount of time discussing plans for next year's legislative effort with the Florida Chapter's lobbyist. Chapter programming is developing and will have the full support of the National Association.

The Scientific Advisory Board is currently evaluating several proposals for research on the syndrome. One of these proposals can lead to the improved ability to diagnose PWS. All of these projects can benefit those we serve.

The mission of the Association is daily being fulfilled by each one of you. As you tell people about the syndrome, support others in the PWSA (USA) family, or advocate for services, you move the cause forward. Remember that in 1975 we began the first and only organization concerned with PWS; now in only 19 years there are organizations all across the country and around the world. Together, there is nothing we cannot do.

Letters to the Editor

Dear PWSA (USA),

I have just read the 1993 September-October issue of your newsletter, *The Gathered View*, for the first time and I have to admit it was the most interesting and informative newsletter of any organization that I've read in a long time.

I am an employee of a human service organization that provides a number of services for people with mental retardation. A number of our participants have PWS and I believe that an information source such as your newsletter gives insight and encouragement to those of us who are responsible for helping care for and nurture the development of people with this syndrome.

The editors, contributing writers and the members of your organization deserve much praise. Your work is not in vain.

Sincerely,
Daniel Harris, Program Specialist
Chimes Metro Inc., Baltimore, MD

Editor's Note: *The Gathered View* is one of the primary means of communication among members of PWSA (USA). We welcome input from all our members. Please send your letters or articles for the newsletter to: Editor, PWSA (USA), 2510 S. Brentwood Blvd., Suite 220, St. Louis, MO 63144-2326, or FAX to 314-962-7869. Comments, suggestions, and questions also can be phoned in at any time: 800-926-4797 or 314-962-7644.

President's Message

Links in the Worldwide Bond

by Janalee Tomaseski-Heinemann

I recently received correspondence from Israel written by a physician who has a 7-1/2-year-old child with Prader-Willi syndrome. She was requesting advice on weight management for her child. In her first fax, she referred to my chapter in the book, *Management of PWS*, edited by Greenswag and Alexander. She states, "...we read and reread your article. We must commend you on the deep perceptions and constructive, yet down to earth, advice you have provided." It wasn't until I received that fax that I realized how far-reaching our Association has become over the years. It is, of course, primarily thanks to Louise Greenswag, Randy Alexander, and their volunteer writers that this book has become so widely read and respected. It is also thanks to PWSA (USA) for financially backing this book and making it a reality.

I have quoted the statistics before that PWSA (USA) has members from 18

countries, but that doesn't really have an impact until you read such letters as those we recently received from Australia and New Zealand, and you see the responses to our sibling questionnaire that came from Italy and South Africa. Our common bond spans all continents and all races. Our common problems and issues strengthen the links of this bond. For example, I'm sure many siblings can relate to the statement from the sibling in South Africa who wrote, "When I was younger--especially during my teens--I was extremely embarrassed about my brother. Even now, I struggle with the embarrassment, especially with those who do not understand the syndrome."

Another important link in our international bond is the International Prader-Willi Organisation, which was formed in 1991 as a "consolidating body for all individual PWS associations throughout the world". IPWSO was founded as the result of the international conference in

Holland. Dr. Suzanne Cassidy, who serves on both the PWSA (USA) Scientific Advisory Board and Board of Directors, played a major role in establishing this first international conference. She is the United States' medical representative for IPWSO and I am the parent delegate. There is both a parent newsletter and a scientific newsletter produced by IPWSO. A second international conference will be held in Norway, June 15-18, 1995.

A new electronic informational network that hopes to be international in scope is being generated by Mr. Shelly Tarakan, publisher of *Prader-Willi Perspectives*. We wish him well in this endeavor. We at PWSA (USA) and IPWSO do not contend to be the only source of information you will ever need, but strive to be an important link in the chain of assistance from family to family, from state to state, and from country to country.

*Help thy brother's boat across,
and lo!
thine own has reached the shore.
--Hindu proverb*

Georgia on Your Mind?

by Dottie Cooper
1994 Conference Chair

As you begin making plans to attend the 1994 National Conference in Georgia this July, you may want to allow some extra time to take in all there is to see and do in the "Peach" state, also known as the "Gateway to the South."

In downtown Atlanta, the Coca-Cola Museum and Underground Atlanta are featured attractions. Our Atlanta Braves baseball team (National League Champions) is really "hot". The fabulous Atlanta Fox Theater (where "Gone with the Wind" premiered) is hosting "Seven Brides for Seven Brothers" all through the week. Don't miss the Martin Luther King Center and the Carter Presidential Center! Of course, golf and fine restaurants are in abundance.

Outside the perimeter of the city is Stone Mountain (east), with its daytime attractions and evening laser show. This family affair covers 3,200 acres of entertainment, recreation, and natural beauty. Enjoy Six Flags over Georgia (west) or boating at Lake Lanier Islands (north).

North of Atlanta, you'll enter the cooler foothills of the Blue Ridge Mountains and find many varieties of arts and crafts. The Highlands (where Georgia, North Carolina, and South Carolina meet) are a favorite vacation spot of Florida residents. Just 90 minutes south of Atlanta, enjoy the beautiful Calloway Gardens. Go a little further and visit Colonial Savannah, Okefenokee Swamp, Fort Walton, and Panama City beaches. East of our city is Hilton Head, Jekyll, and St. Simons Islands where you will find beaches and golf courses. Traveling west, you'll find the famous Boaz Outlet and Huntsville Space Center in Alabama. Chattanooga and Nashville, Tennessee are just north.

Yes, there's lots to do, so plan some extra time before and after the conference to make the trip a family vacation you'll always remember. For more information and a free Georgia map, call the Georgia Department of Industry, Trade, and Tourism at 1-800-847-4842 and ask for the "1994 Georgia on My Mind" catalog.

"Georgia on My Mind"

PWSA (USA) National Conference

Atlanta, Georgia - July 21-23, 1994

The months leading up to the PWSA (USA) National Conference to be hosted in Atlanta, July 21-23, 1994, are quickly passing by, and the Georgia Chapter has been busily putting together the details. We think we have agendas for both the adult and youth programs that will be new, exciting, informative, and fun. Our theme for the conference is none other than "Georgia on My Mind." That's what we think will be on the minds of families, friends, and professionals interested in Prader-Willi across the nation this July.

For those of you who have never attended a Prader-Willi Syndrome Association (USA) National Conference, this is an opportunity to meet and learn from other families and professionals from all over the country who truly understand Prader-Willi syndrome. We hope that those who are returning will enjoy seeing old friends and making new ones, and participating in exciting new programs tailored to your needs.

In the true spirit of "Southern Hospitality," some of the major corporations in Atlanta are supporting our

national conference through corporate sponsorship. Contributions are continuing to roll in. Thus far we have received contributions from the following:

- * BellSouth Telecommunications
- * Lotus Corporation
- * Westinghouse Corporation
- * Amoco Corporation
- * The Georgia Governor's Council on Developmental Disabilities

Monies and "in kind" donations received will be used to enhance and supplement conference expenses. Donations from our corporate sponsors allow us to maintain the low cost and high quality of programs organized for attendees. We very much appreciate their support.

Registration materials have been mailed to our membership. Notify the National Office to receive additional packets, 800-926-4797. We look forward to one of the largest conferences in the history of PWSA (USA).

Adult Program Special Features:

- * *Special welcome session for "newcomers." Social session for "old timers."*
- * *Dental presentation by Dr. Kari Storhaug, Norway.*
- * *Presentations targeted to interest, based on age of person w/ Prader-Willi: (0-5), (6-11), (12-17), (18+).*
- * *Popular special sessions "For Mothers Only," "For Fathers Only," and new, "For Siblings Only."*
- * *Unveiling of the new PWSA (USA) logo. Meet the National Office staff.*
- * *Introduction to PW Syndrome Awareness Day.*
- * *Standard favorites; Overview of PWS, updates on Scientific Conference, Residential Providers' Workshop, and Presidents' Day.*
- * *Coffee/dessert social, banquet/dance, entertainment...*

16th Annual National Conference

Pre-Conference, July 20

- * Scientific Conference (Prof. only)
- * Residential Providers Workshop
- * Chapter Presidents Meeting
- * Conference Registration Opens

Thursday-Saturday, July 21-23

- * Conference Sessions
- * General Membership Meetings
- * Social Events
- * Youth/Adult Activity Program (YAAP)

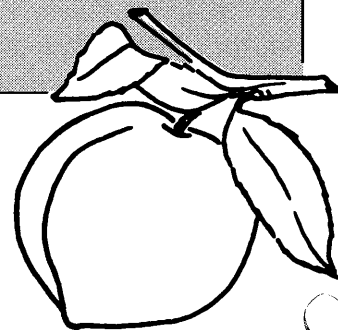
Atlanta Marriott Northwest

I-75 at Windy Hill Road
200 Interstate North Parkway
Atlanta, Georgia 30339
404-952-7900

To request conference packets, call:
800-926-4797 or 314-962-7644

FUN! FUN! FUN!

Youth/Adult Activity Program (YAAP) for those with PWS and their siblings, lots of exciting things to see and do! (Featured in January/February '94 *Gathered View*).



— Local/International News —

While the National Office has been busy with its move to St. Louis, the rest of the country has been far from idle. Chapters have been busy with plans, meetings, and activities. Statewide and regional conferences are mushrooming, social events abound, progress toward or achievement of group homes is happening (albeit slowly). Whether the groups are large or small, there is a marvelous sense of vitality, support and camaraderie. Here are just a few of the "goings-on":

Steve Lundh has taken on the daunting task of being Chairman for the 1995 PWSA (USA) National Conference, to be hosted by the **Prader-Willi Northwest Association**. Steve and his committee have chosen Double Tree Suites Hotel in Seattle, Washington as the site.

Prader-Willi Florida Association publicized lots of exciting plans in their "Gator-Willi" newsletter for their all-day Spring Conference in Orlando on April 23rd. Featured speaker: Dr. Patrick McGreevy, who currently serves as Behavior Analyst for Exceptional Student Education in the Orange County Public Schools in Orlando.

Like to travel? The March 1994 newsletter of PWSA (UK) reported on their 1993 annual conference in England, which 252 people attended, as well as the upcoming 1994 annual conference October 29-30. Should you be considering a trip to Australia, the newsletter of the **PWSA of Australia** announced their 5th national conference on October 1-2, 1994, "Focussing on Families," in Hahndorf, South Australia. The family of PWS truly stretches around the world! (Interesting note: the oldest known person with PWS in the UK died at the ripe old age of 71!)

A one-day conference in Langhorne, Pennsylvania, on "Alternative Living Options for Individuals with PWS" was sponsored by **PWSA of Pennsylvania** on April 9th. Of additional interest was testing of children there by the Monell Chemical Senses Center to investigate how food preferences of children with PWS shift with changes in sweetness, saltiness, bitterness, and fat content of food; their sense of smell was also tested.

PWSA of North Carolina rejoices! After lots of hard work, the home for people with PWS was approved by HUD and will, it is hoped, be ready to start serving six persons in mid-1995.

PWSA of Missouri held their annual Bowl-A-Thon to raise money to help youth/adults with PWS attend Wonderland Camp located near Lake of the Ozarks, Missouri. Parents, relatives, friends and children had a great time bowling!

Conference Grants Available

How do you apply for a conference grant?

Send PWSA (USA) a letter which includes:

1. the size of your family, and age of your child(ren);
2. an indication of your income and expenses;
3. a brief summary of difficulties your family is experiencing in dealing with PWS;
4. whether your family would need all your conference expenses paid or if a partial grant would be sufficient (e.g., for travel or lodging only);
5. whether anyone in your family has ever attended a PWSA national conference.

Calendar of Events

To publicize your group's events in the May/June Gathered View, send in dates by May 25th to the National Office.

May 7th -PWSA (Ohio) meeting, 9:00 a.m. Mildred Lacy, speaker, from PWSA (Kentucky) and member of PWSA (USA) Board of Directors. Piano Room, Children's Hospital Medical Ctr., Cincinnati, Ohio. Contact: James Boyle, 216-932-3587 or 216-741-6778

May 15th -PWSA (Maryland) meeting, 1:30 p.m. Beltsville, Maryland, Contact: Linda Keder, 301-384-4955

June 18th -PWSA (Michigan) meeting, 10:30 a.m. East Lansing Public Library, Contact: Helen Warner-Bell, 313-626-8995

June 25th -PWSA (Northwest) Annual PWNA Picnic, 1:00 p.m., McSwan's Farm - Pasco, Contact: Angus & Nancy MacDonald, 206-285-7679

PWSA (USA) offers grants to families wishing to attend the national conference but whose finances prohibit this expense.

What is the process?

Your letter will be reviewed by the conference grants committee. Letters requesting conference grants must be received at the National Office by Friday, May 25, 1994. Applications received after this date cannot be considered.

Hormones and the Hypothalamus

by Adrian D. Sandler, M.D., Program Director, Neurodevelopmental Nutrition Program, University of North Carolina at Chapel Hill

We are so used to hearing about the storm and stress of adolescence, when sexual interest, acne and family conflict all escalate together, that we sometimes forget the importance of hormones in the pre-adolescent years. Certainly it is true that puberty is intimately related to hormonal surges (which are blunted in teenagers with Prader-Willi syndrome), but those same hormones are playing important roles even before birth. The medical science of hormones is endocrinology.

In order to understand some of the complex endocrinological aspects of Prader-Willi syndrome, it is necessary to know something about the *hypothalamic-pituitary axis*.

The hypothalamus and pituitary gland are vital structures in the brain that act like the conductors of the endocrine orchestra. The hypothalamus (an immensely interesting part of the brain which among other things regulates sleep, appetite, growth, and temperature) secretes peptide releasing factors (RF) which stimulate the pituitary gland beneath it. Specific cells in the pituitary secrete specific hormones in response to this stimulation. For example, *luteinizing hormone releasing factor* (LRF) stimulates release of certain pituitary hormones, *luteinizing hormone* (LH) and *follicle stimulating hormone* (FSH). LH and FSH in turn travel in the circulation to the testes and ovaries where they stimulate the secretion of the sex hormones testosterone, progesterone and estrogen. These hormones are primarily responsible for de-

velopment of the genital organs, breast development, menstruation, and aspects of sexual behavior. The system tends to regulate itself with elaborate feedback mechanisms, which keep things in balance, while at the same time allowing cyclical surges and major readjustments during puberty and at other stages in development.

In the same way, the hypothalamus and pituitary gland regulate thyroid hormones, growth hormones, stress hormones, and hormones regulating metabolism and salt and water balance.

So many aspects of PWS are clearly related to these hormone systems that the hypothalamus is undoubtedly the primary area of dysfunction.

Let us consider hypogonadism or incomplete development of the sexual organs. Hypogonadism is usually evident at birth in male infants with PWS, who frequently have a small penis, undescended testes, and underdeveloped scrotum. Hypogonadism may be present but less obvious in baby girls with PWS. During adolescence, sexual maturation is usually (but not always) delayed, atypical, or incomplete. Many girls may have normal early breast development, but then other puberty changes, including the development of the areola, nipple, labia

minora, and clitoris may be delayed or incomplete. Most will have somewhat delayed onset of periods at around 11 to 16 years of age, and their periods may be irregular and infrequent. Most boys have delayed onset of puberty, so that penis and testes remain small during the early teen years, and there is little change in voice or body hair. Some males continue to have gradual sexual development during the later teen years, however. Although I have seen adult males and females with PWS who appear to be quite well developed sexually, there have been no documented cases of fertility, to my knowledge.

Careful study by endocrinologists has demonstrated that hypogonadism in PWS is secondary to a deficiency of LRF in the hypothalamus. This must be present as early as the second trimester of pregnancy to account for the hypogonadism noted at birth. Blood levels of testosterone and estrogen are low in adolescents and adults with PWS, and the secretion of LH and FSH in response to injection of LRF is blunted. Abnormal growth hormone secretion and stimulation in many children with PWS also implicates the hypothalamic-pituitary axis. I have also seen a few individuals with PWS who have borderline low thyroid hormone values, presumably from the same underlying cause. In addition, the behavioral characteristics of PWS are likely to be related to hypothalamic dysfunction. Hunger, food-seeking, insatiability, excessive sleepiness and hypotonia, lethargy, high pain threshold and skin-picking are also related to the hypothalamus.

What are the implications for treatment?

First, I think that the majority of individuals with PWS can benefit from hormone therapy, but the decisions must be made on an individual basis.

The treatment of an abnormally small penis is simple enough and usually effective, and so I recommend this treatment if the condition is likely to be noticeable in the future by peers in showers and changing rooms. Three doses of intramuscular Depo-Testosterone 25 mg one month apart are sufficient and unlikely to cause side effects such as persistent erections. It should be done before 3 or 4 years of age to prevent making the child sensitive about the size of his penis.

Undescended testes can sometimes be made to descend into the scrotum with shots of a hormone hCG (3,300 IU/week for 4 weeks), but this is seldom successful if the testes are very high in the inguinal canal or still in the abdomen. Occasionally, the testes may descend spontaneously during teen years in PWS, and so it may be worth waiting until then before having surgery to try to bring the testes down.

The use of testosterone in the teen years to advance pubertal development in males is controversial, and there is no published research to guide treatment decisions. Although some centers such as University of Connecticut Health Center have used Depo-Testosterone in many adolescent males with PWS, I have no personal experience in this to date. There are justifiable concerns about aggressive behavior and advancement of bone age, and it is clear that such treatment should be closely monitored by a pediatric endocrinologist, preferably as part of a research study.

On the other hand, I have had some experience in the use of long-term *estrogen therapy in females with PWS*. The simplest treatment is with the oral contraceptive pill. Potential complications include blood clotting and high blood pressure, and so it is important to control obesity before beginning therapy and to

monitor therapy closely (including regular gynecological exams). The advantages of such treatment include the psychological benefits of enhanced pubertal development and regular and predictable menstrual periods. Another very important potential advantage is that estrogen therapy can enhance bone mineralization. Most adults with PWS have significant osteoporosis (brittle bones) which can lead to fractures and spinal deformities. The use of estrogen in young women may be a valuable preventive health measure. Similarly, the use of testosterone in males may help to prevent osteoporosis. I would also recommend regular weight-bearing exercise and checking that the diet is adequate in the intake of vitamin D and calcium to keep those bones strong!

(Editor's Note: This article is reprinted from the newsletter of the Prader-Willi Syndrome Association of North Carolina, with the permission of Dr. Sandler. Italics were added.)

Research Projects / Clinical Services

NDNP Offers Assessment and Care

The Neurodevelopmental Nutrition Program (NDNP) is a joint program of the University of North Carolina's Clinical Center for the Study of Development and Learning, Genetic Counseling Program, and Department of Pediatrics, and the Prader-Willi Syndrome Association of North Carolina (a PWSA chapter). Dr. Adrian Sandler, who wrote the accompanying article on hormones and the hypothalamus, is a pediatrician and program director at the NDNP.

NDNP offers interdisciplinary assessment and family-focused care for infants, children, adolescents, and young adults with problems of development and nutrition. These include Prader-Willi syndrome, oral-motor problems, rumination, feeding disorders, and developmental disabilities accompanied by obesity. While the program is not exclusively for those with PWS, it was established with PWS in mind.

Comprehensive diagnostics, treatment, health maintenance, and service coordination are provided by a professional group that includes genetics, dentistry, nutrition, pediatrics, physical therapy, psychology, and speech and language pathology. Clinics are held on the first Monday of every month and include group discussions and health education as well as individual services.

UNC's program, in conjunction with the Prader-Willi Syndrome Association of NC, also serves as a focal point for dissemination of information about PWS to the state's health care community.

For more information about the program or to make a referral, contact: Kay Hains, Asst. Dir., Clinical Services: (919) 966-5171.

Families Needed for Behavior Research

A new study proposed by Barbara Whitman, Ph.D., seeks to determine if extended family health histories (often called family pedigrees) can help predict those persons with Prader-Willi syndrome who are more at risk for serious behavior difficulties. If, by looking at such family histories we are able to inform parents early on of increased risk for behavior problems, we may also be able to institute measures to avert these problems. To investigate these questions, parents or caregivers will be asked to complete a behavior checklist regarding their persons with Prader-Willi syndrome, construct a "health family tree," and complete two measures of current family stress and ways of solving problems. The role of family history and current stress will be looked at in relation to behavior and emotional dimensions.

This research is a natural next step to our previous 10 year's work into the behavioral and emotional dimensions of Prader-Willi syndrome and how best to manage these dimensions - both behaviorally and with medication. After discussion with many professionals at last summer's conference, this research was formulated and initiated April 10, 1994 at the New England Prader-Willi Association meeting. Our previous work into behaviors and medications has been widely disseminated among members of PWSA for their use. This information will also be published for all to be able to use. Data, however, will be compiled in group form so that no one individual can be identified.

For more information or to participate in this research project, please contact: Barbara Whitman, Ph.D., St. Louis University Department of Pediatrics, 1465 S. Grand, St. Louis, MO 63104, 314-577-5609

Two Paths Lead to the Same School

Adam Sporbert of Southington, Connecticut, and Kimberly Pacheco of West Hartford, Connecticut, have a couple of things in common. Both of these children were placed by their towns in the Intensive Education Center (IEC) in West Hartford, and they both have Prader-Willi syndrome. Adam is 10 and Kimberly is 7, and they are in the same class. It is very rare to see two children with PWS in the same classroom, especially when the program isn't specifically for children with PWS.

The IEC is a private, special-needs school. It wasn't until last year that the staff there ever heard of Prader-Willi syndrome. The director of the school, Sister Helen Dowd, became very interested in PWS and attended the Philadelphia [PWSA national] conference prior to the start of Adam's school year, in 1992. It was at this conference that the Pachecos met Sister Helen Dowd and were told about the school. The Pachecos were from the same town and weren't even aware of the school's existence, as it is very small -- about 30 students.

He was welcomed with open arms.

In September 1992, Adam started at IEC. He was welcomed with open arms. The staff are truly concerned about his diet and welfare. His counseling sessions are done while taking a brisk walk rather than sitting in a room. He is sent on errands to give him added opportunity for movement. The staff is constantly revising the program to meet his needs, especially when they receive new literature with ideas on behavioral techniques.

That same September Kimberly went back to school, but to public school. There were problems with her aggressive behavior in the classroom. The school continually used time-out until it no longer served a purpose. They then asked the parents to pick the child up from school when she was aggressive, thinking this would be the solution. The problems escalated, and the parents were called more and more frequently, until there was very little learning taking place. Her skin-picking had increased, and she was becoming a very unhappy and difficult child.

In February of 1993, Kimberly was placed at IEC at the request of her parents. Immediately the skin-picking began to improve and clear up -- a sign that she was experiencing less stress. Her aggression decreased and this year is almost nonexistent.

For both Adam and Kim one thing is the same -- they feel good about themselves. The school prides itself on having all children have high self-esteem and helping each child reach his or her highest potential. Adam and Kim are proud of the work that they do, and they are eager to learn new things. They have achieved so much in such a short time.

As parents, we feel there couldn't be a better placement for our children. These children share a common bond -- PWS -- and are starting to develop a special friendship with each other. They report to us each day and let us parents know how the other child is doing. We are proof that a proper placement can make a difference.

*--Barbara Pacheco and Barbara Sporbert
(parents of Kim and Adam)*

.....

Summer Camp:

For any parent, the decision to send their child to summer resident camp is made with careful consideration. It is equally important to choose the right camp with the appropriate activities and programs to meet the special needs of a person with Prader-Willi syndrome. Some hints when choosing a summer camp -

- * **Administration and Camp Staff:** Check to see if all camp personnel are knowledgeable and trained concerning Prader-Willi syndrome. Ask to speak to the Camp Director or Program Director; they should be happy to answer your questions and make you feel at ease.
- * **Safety and security:** Ask to tour the campgrounds and check the facilities. Ask about camper/staff ratios, qualifications for staff employment, security and health personnel, food security, and discipline/behavior control.
- * **Program Content:** Summer camp should be a happy, fun experience for any camper! Building self-esteem, success, and confidence within each individual must be a goal. Ask if campers are grouped according to age or functioning level within each activity. Learning new things and developing social skills are an important part of any camp experience.
- * **Accredited or sponsored by a well-known, organization /sponsor.** The American Camping Association accredits camps that meet rigorous educational and recreational standards in four areas: administration, program, personnel, and campsite. Major sponsors or organizations support specific camps on the basis of excellence/image in the community.

Editor's Note: The Missouri Chapter of PWSA (USA) has made arrangements with Wonderland Camp, located on the Lake of the Ozarks in Missouri, to have an all PW week of summer camp, June 26-July 2. A minimum of 55 campers with PWS needs to be registered by the end of May. If this number is not reached, other campers with various handicaps may attend. For more information or applications, contact as soon as possible: Cari Williams or Alan Moore at Wonderland Camp (314) 392-1000.

Diagnosis Doesn't Equal "Doom and Gloom"

by Marge Wett

Many parents reading about PWS for the first time state that what they read is all "doom and gloom" or use such adjectives as "devastating." New parents find reality hard, a situation we wish we could change. The fact is PWS is a rather complex set of symptoms that will affect this infant. You have lost that "normal" child you expected, and that is not easy to face.

Through years of experience in working with infants, children, adults, parents, and professionals, I used the phrase "the sunshine kids." I don't know if that phrase is original, but I do frequently use it when referring to the people with PWS who do not exhibit some of the more bizarre behaviors you have all read about. I believe we need to read about success too, in order to balance our perspectives.

Even with your child's handicaps, you do have a child that can achieve the goals you want for your children. My list of goals includes happiness, reasonably good health, and achievements. My daughter Lisa, who has PWS, has accomplished everything on my list. Some people describe Lisa as having a "mild" case of PWS. To me that was as if saying I have a "mild" case of pregnancy. Having been through seven pregnancies, I can say they are not all alike. PWS is not the same in every case either.

Lisa's diagnosis came in her teens, but we worked with the symptoms from the day she was born. The hard reality for us was leaving her in the hospital for the

first three weeks of her life and then taking home the very flaccid baby, who required gavage feeding. It was hard reality for my husband, Dick, [a physician] when she returned to the hospital at five weeks for some tests and he was the one who responded to the emergency call when she reacted to the testing and we almost lost her. The hardest part of the testing was it gave us no answers, and later I would find myself telling parents they were fortunate to receive an earlier diagnosis although they couldn't always see that.

Lisa's needs were different from our other children's, and frequently more difficult, but I never had the "doom and gloom" attitude. It's true Lisa is one of the "sunshine kids," but we had our share of facing PW problems too. We survived Lisa's problems just as we survived our other children's problems and their joys.

Like our other children, time came for Lisa to move away from home. Moving from our home into a group home situation was a matter of meeting her needs, and it has continued to over the years. Her home has 15 residents, which is a "no-no" in current providers' thinking, but it is one of the most successful homes I have visited. She is quite satisfied with her living arrangements, her high number of outside leisure activities, and her work. She works in a sheltered workshop two days a week and in the office of an HMO three days a week. She looks forward to a weekend a month and special trips for family celebrations and holidays, but she goes "home" just like our other children. She just celebrated her 29th birthday in March and enjoyed taking two male friends out to dinner and to a movie.

When your child is young, I believe in facing the reality of the syndrome. One of those realities is that your child can accomplish goals despite their "differences," even if they aren't one of the "sunshine kids."

Medical News

Gene Therapy Proves Successful for One Genetic Disease

In the first published account of a successful gene therapy treatment, scientists at the University of Pennsylvania report that a woman with a rare and deadly hereditary disease has seen significant improvement since a gene correction treatment nearly two years ago.

The patient, a 30-year-old Canadian woman, suffers from familial hypercholesterolemia, which causes cholesterol levels to soar eight to 10 times above normal, usually resulting in death from heart disease by young adulthood. The treatment involved removing a portion of the woman's liver, exposing the liver cells to a virus that contained a cholesterol-lowering gene found lacking in those with the disease, and reinfusing the improved cells directly into the liver. While not a complete cure, the treatment significantly reduced the patient's cholesterol levels and halted the advance of the disease. Results of the experimental treatment are detailed in the March 31 issue of the journal *Nature Genetics*.

Although treatments or cures for other genetic disorders, including Prader-Willi syndrome, may be a long way off or even impossible, this case is viewed by many as a promising landmark. According to Francis Collins, director of the National Center for Human Genome Research at the National Institutes of Health, "This is proof that this approach, which has been talked about so much and has stirred a bit of controversy, can do what it's supposed to do." Says Collins: "Gene therapy is still very much in its early stages, but a few decades from now, when people look back, they'll see this as a significant milestone."

Editor's Note: Marge Wett was one of the founding parents of PWSA, served as its executive director for 12 years, and has contributed countless volunteer hours to the Association over the years. We thank Marge for her continued involvement in submitting this article for publication.

Question Box

Q:

At age 5, my child still wears a diaper at night and is on the verge of outgrowing the largest size of diapers and Pull-Ups readily available. I'm beginning to wonder how long this incontinence might go on, given her PWS, and whether we should consider some medical or behavioral strategies at some point in the future. Do you have any advice on night-time training, and do you know of any good sources of larger diapers or training pants that would be appropriate for her in the meantime?

(This question is answered by Dr. Suzanne Cassidy, Professor of Genetics and Pediatrics at Case Western Reserve University and member of PWSA's Board of Directors and Scientific Advisory Board.)

A: Nighttime incontinence, or enuresis, is extremely common in the general population and may last up to age 7 without requiring evaluation, assuming that there is no daytime incontinence. It is often found that one of the parents of a child who has enuresis also had enuresis as a child. In Prader-Willi syndrome, or in any other disorder in which there is delayed developmental maturation, the age at which one no longer sees enuresis may be even older than 7 years and may still not indicate a medical problem. However, at about age 7 it is appropriate to see a urologist for an evaluation.

You used the term "incontinence" rather than "enuresis." Incontinence usually implies that there is no control of urination and that it occurs throughout the night, whereas enuresis implies one or two identifiable events of loss of urine. If incontinence is truly present, a medical evaluation is appropriate. It is enuresis which is common and usually benign.

There are many strategies for trying to decrease enuresis. The most straightforward is to have the child drink no fluids after dinner and to awaken the child to urinate at the time that the parents go to bed. Behavior modification techniques by rewarding the child for not wetting the bed are also of use in some cases in decreasing the frequency with which enuresis occurs. There is a medication called DDAVP, which is a synthetic version of a hormone normally secreted by the pituitary gland called the antidiuretic hormone. Its function is to stop elimination of liquid. DDAVP is usually taken by inhalation in the nose, and can be quite effective in treating enuresis that does not have an organic basis.

You inquire about larger diapers. Perhaps this is a question for your pediatrician. Have you tried Depends or some of the other diapers intended for older adults, who also have problems with incontinence?

In summary, urinating once or twice during the night is quite common in the general population and even more common in disorders with developmental delay, such as Prader-Willi syndrome. Restriction of fluids in the evening, awakening to urinate, and behavioral encouragement for dry nights are the first approach. If these are unsuccessful, medication might be tried by the pediatrician. A urologist is the appropriate physician to evaluate for structural or other biological causes of enuresis after age 7.

Editor's Note: At least two manufacturers of baby diapers (Procter & Gamble and Kimberly Clark) also make disposable diapers in youth and adult sizes. If you're unable to find their products (Depends and Attends) locally, or if they don't meet your child's needs, you can order a wide range of either disposable or reusable diaper products from a number of manufacturers and distributors. Many have free catalogs and mail delivery service. (See list at right.)

If you prefer to use a washable product, you might try Nikky brand absorbent pants or similar products found in some baby products catalogs. Other sources of reusable products in larger sizes include Duraline (see list) and TOP DRAWERS, 901 Mainstreet, Suite A, Hopkins, MN 55343. For other products and suppliers, you may wish to contact Help for Incontinent People, Inc. (HIP), Box 544, Union, SC 29379, Telephone 1-800-BLADDER. HIP compiles and sells a Resource Guide of Continence Products and Services as well as publishing a quarterly newsletter.

Although we cannot personally recommend any specific products, here are some sources of disposables you might try:

Access Medical Supply
2006 Crown Plaza Drive
Columbus, OH 43235
(800) 242-2460

CompCare
5534 Cortez Road
WestBradenton, FL 34210
(800) 877-7317

Duraline Medical Products
324 Werner St.
Leipsic, OH 45856
(800) 654-3376

HDIS
325 Paul Ave.
Ferguson, MO 63135
(800) 538-1036

Woodbury Products, Inc.
4410 Austin Boulevard, Dept. 250
Island Park, NY 11558
(800) 777-1111

Q:

Realistically, can we expect our child with PWS to carry his share of chores around the house?

(The following answer is from Bea Maier, Senior Coordinator at The Rehabilitation Institute of Pittsburgh.)

A: Doing chores is a normal part of life. It helps a child's self esteem to be a contributing member of the family, and it is perfectly normal for kids to grumble about taking out the garbage. It is

The Parent-Professional Relationship:

Advice From Experienced Parents

(The following is an excerpt from "Parenting A Child With Special Needs: A Guide to Readings and Resources," NICHCY News Digest Vol. III, No. 1, 1993, The National Information Center for Children and Youth with Disabilities)

There should be a mutuality in the parent/professional relationship. Both parents and professionals need to trust and feel trusted, both need to admit when they do not know or are wrong, and both need to negotiate with each other. Trust, respect, and open communication between parent and professional are, therefore, essential to building a good working relationship. This can take time to develop and may require effort from both parties. To that end, many parent writers suggest:

A: Chores, *Continued from p. 10*

not a kindness to cut them out of any family activity, and protest and grumbling are a part of it. All children learn there are obligations—moms have to make dinner, parents go to work. This is one way of teaching responsibility.

Start early. A child who is walking and has relatively good use of arms and hands can do a simple chore like, say, fetching a towel. Another example is that young children can learn to put their clothes in a laundry hamper. Initially they won't remember, and it will require prompting and cues from the adult. Early training and realistic expectations help develop social skills and attitudes, regardless of developmental level. This orientation may mean more work for Mom and Dad, but it will pay off later.

- * If you are looking for a specialist with whom you can work well, ask other parents of children with disabilities. Often, they can suggest the name of a good speech or physical therapist, doctor, dentist, surgeon, and so on.

- * If you don't understand the terminology a professional uses, ask questions. Say, "What do you mean by that? We don't understand."

- * If necessary, write down the professional's answers. This is particularly useful in medical situations when a medication or therapy is to be administered.

- * Learn as much as you can about your child's disability. This will assist you with your child, and it can help you participate most fully in the team process.

- * Prepare for visits to the doctor, therapist, or school by writing down a list of the questions or concerns you would like to discuss with the professional.

- * Keep a notebook in which you write down information concerning your special needs child. This can include your child's medical history, test results, observations about behavior or symptoms that will help the professional do his or her job, and so on. (A loose-leaf notebook is easy to maintain and add information to.)

- * If you don't agree with a professional's recommendations, say so. Be as specific as you can about why you don't agree.

- * Do whatever informed "shopping around" and "doctor-hopping" are necessary to feel certain you have explored every possibility and potential. As Irving Dickman [author of *One Miracle at a Time*] says, "Shop. Hop. Hope."

- * Measure a professional's recommendations for home treatment programs or other interventions against your own schedule, finances, and other commitments. You may not be able to follow all advice or take on one more thing, feeling as Helen Featherstone [author of *A Difference in the Family*] did when she wrote, "What am I supposed to give up? ... There is no time in my life that hasn't been spoken for, and for every 15-minute activity that has been added, one has to be taken away." Peggy Finston [author of *Parenting Plus*] points out that "most professionals won't be familiar with the sum total of our obligations and will not take it upon themselves to give us permission to quit. This is up to us. It's in our power to make the decision."

Editor's Note: For a free copy of the 23-page News Digest, which includes articles, references, and resource lists, contact NICHCY, P.O. Box 1492, Washington, DC 20013, Telephone 1-800-695-0285 (or 202-416-0300 in the D.C. metro area). "A Parent's Guide to Doctors, Disabilities, and the Family" also is available free of charge from NICHCY, as are many other helpful publications.

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Nominations to PWSA (USA) Board of Directors

The Nominating Committee requests that the names of members interested in, or recommended for, a seat on the PWSA (USA) Board of Directors be submitted to the committee no later than May 25th. Recommendations for nominees, along with a brief description of person's qualifications, should be sent to the attention of:

Annette Ruiz, Nominating Committee Chair
c/o PWSA (USA), 2510 S. Brentwood Blvd., Suite 220,
St. Louis, MO 63144-2326, FAX 1-314-962-7869.

Research Project Update

Editor's Note: Last fall the *Gathered View* published news of a proposed Prader-Willi Syndrome Research Project at Vanderbilt University School of Medicine in Nashville, Tennessee, which had been submitted to the National Institutes of Health for funding, and asked PWSA (USA) members to support it by writing to NIH. Following is an update on its current status from Merlin G. Butler, M.D., Ph.D.

Our project was not funded last year; however, we have resubmitted our grant application (modified slightly) for further consideration for funding. We should know during the next several months whether this grant application has been accepted for funding. We think the grant application is worthy of support, and we look forward to working with Prader-Willi syndrome families to gain a better understanding of the cause and treatment of individuals with this condition.

The Gathered View is the official newsletter of the Prader-Willi Syndrome Association and is sent to all members. The opinions expressed in *The Gathered View* represent those of the authors of the articles published, and do not necessarily reflect the opinion or position of the officers and Board of Directors of PWSA (USA). Duplication of this newsletter for distribution is prohibited. Quotations may be used if credit is given to PWSA (USA). Annual membership dues: \$21 Individual, \$26 Family, \$31 Agencies/Professionals (U.S. Funds). Send dues, change of address, and letters to: PWSA(USA), 2510 S. Brentwood Blvd., Suite 220, St. Louis, MO 63144-2326. Any questions? Call: 800-926-4797 or 314-962-7644 or FAX 314-962-7869.