The Gathered View

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

Vanderbilt Proposes Major PWS Study

An interdisciplinary team from Vanderbilt University, which has been conducting research on Prader-Willi syndrome, now is seeking federal funds to expand their work. They propose a comprehensive, interdisciplinary research initiative that would increase understanding of and interventions for PWS, as well as offering benefits to those studying related conditions.

The following summary of the Vanderbilt proposal is provided by Merlin G. Butler, M.D., Ph.D., Associate Professor of Pediatrics and Pathology, ABMG Certified Clinical Geneticist and Cytogeneticist, Vanderbilt University.

In order to address the main genetic and behavior issues in Prader-Willi syndrome (PWS) patients, Vanderbilt's Kennedy Center and Departments of Pediatrics, Orthopedics, Pathology, Neurology, Radiology, Medicine, and Psychiatry have developed a research protocol to characterize definitively the clinical features of patients with PWS as they relate to chromosomal and DNA identified typologies and to investigate specific behavioral, perceptual, neuroanatomical, neuroendocrine, nutritional, metabolic and physiological features that may distinguish genetic/chromosomal subtypes of PWS. This proposal encompasses the most detailed and extensive study to date of people with PWS.

Types of Testing Proposed

A battery of psychobehavioral, clinical and laboratory tests will be conducted including:

- construction of a genetic map of chromosome 15;
- cognitive and affective assessments;
- ecobehavioral observations in the natural environment;
- visual processing and ophthalmological assessments;
- neuropharmacological intervention;
- radiological examination of bone (e.g., for osteoporosis);
- brain imaging (positron emission tomography, magnetic resonance imaging);
- neuroendocrine and metabolic tests using high precision liquid chromatography;
- fatness distribution and patterning and body composition determination;
- and nutritional, metabolic and sleep assessments.

Some of these studies require blood samples, while other studies require overnight stay in a metabolic chamber to determine the patient's metabolic rate and overnight stay at our sleep disorder center for EEGs and sleep assessments. Other studies require a battery of behavioral, psychological, and visual tests or brain and other imaging to determine specific central nervous system abnormalities, osteoporosis, or percent body fat, as well as muscle and fat metabolism.

Thus, we plan a detailed and extensive evaluation of Prader-Willi syndrome patients to help characterize features of this disorder, as well as to gain a better understanding of their cause (e.g., genetic, metabolic).

How Results Will Be Used

The data from these shared studies will be entered into a common database, making it possible to explore patterns, differences, or similarities of the foregoing laboratory and clinical tests of persons falling into the two major

continued on page 2

Terms to Know:

Ecobehavioral - behavior in relationship to the environment

Neuroendocrine - pertains to the relationship between the nervous system and endocrine apparatus (e.g., insulin secreting cells, pituitary glands)

Neuroeopharmacological - study of drugs that alter neurological systems

Positron Emission Tomography - (known as a PET Scan) This test makes sectional images taken by a rotating x-ray machine. The x-rays are of local metabolic and physiological functions in tissues. The x-rays trace a natural biochemical substance which is injected into the patient. These traces are the path and speed of photons, which are then tracked by computer. The computer produces a color image and the different colors (or lack of color) are indicators of altered metabolism.

Chromatography - a laboratory technique by which chemicals are separated onto a paper or gel. The rate (fast or slow) at which the chemicals are absorbed allows them to be individually detected. This allows the physician to separate and identify chemicals.

In This Issue . . .

genetic subcategories of PWS (deletion and non-deletion). The data will be useful for descriptive, diagnostic, and prognostic purposes and will be shared with the PWS families. The significance of this comprehensive research initiative for those with Prader-Willi syndrome is that it is designed to lead to therapeutic interventions, including behavior and educational interventions and neuropharmacological treatment.

Broader Applications

Identifying the specific genetic factors responsible for behavior problems such as a ravenous appetite would provide a useful model for studying other developmental behavior disorders, including self-injury and some types of developmental disabilities that have major health care and education complications. While it may not be possible to link all of the given problems to a specific genetic site, it is likely such relationships will be identified for certain developmental disorders (e.g., some types of autism, Rett syndrome, etc.). Identifying neuroendocrine or other metabolic pathways influenced by such a genetic mechanism could have implications for treating related behavior disorders.

Obesity, a cardinal feature in PWS, promotes serious public health problems in the United States, including heart disease, malignancy, hypertension, diabetes, and depression. Increasing incidence of obesity in children and adolescents, coupled with the knowledge that obesity in adolescence tracks into adulthood, indicates that obesity may become an even more serious health problem. One goal of the PWS interdisciplinary research program is to identify genetic and/or metabolic markers that may predict the likelihood of later development of obesity and provide a basis for its prevention.

In summary, there are over a dozen colleagues from several departments at Vanderbilt University that are involved in the study of PWS. Our proposal has been submitted to The National Institutes of Health (NIH) for funding in order to undertake this study on a population of people with PWS from various disciplinary vantage points. This study is ongoing in a limited fashion now but will require additional funding to support the investigations of many of the researchers at Vanderbilt University that have proposed research projects involving PWS.

PWSA members can help!

Please urge NIH to fund this research project by writing to:

Dr. Duane Alexander,
Director NICHD
National Institutes of Health
Bldg. 31, Room 2A03
9000 Rockville Pike
Bethesda, MD 20892

Dr. Butler suggests that letters supporting PWS research addressed to Dr. Alexander may be more forceful if written by PWSA state or regional chapters and signed by members of these chapters. Letters to U.S. Senators or U.S. Representatives would also be appropriate.

Watch The Gathered View for news of the status of this proposal and, if it wins funding, for information on participating in the study.

Credit Card Update

The MBNA MasterCard program continues to provide additional funds for PWSA(USA) programs, support, and publications.

For the first quarter of 1993 we received a check for $190.95. We currently have 111 cardholders, who in the first quarter had 1,134 transactions. Doing our math, this means an average of two to three transactions per person per month.

If we could double the number of people with these cards, we would be providing PWSA with over $1,500 a year, and that is merely using the card two or three times a month.

We encourage our members to use the card for simple transactions such as purchasing your gas. You accumulate a bill that you know you will be able to pay, and you provide support for PWSA.

Another promotion will be coming to everyone soon—please consider participating to support PWSA(USA).

The Prader-Willi Marathon

by Janalee Tomaseski-Heinemann

While our son, Matt, was growing up, many of you followed our Prader-Willi exploits through my personal writings. At that time, I began receiving letters from an Australian mother whose child, Ryley, was just a toddler at the time. Since then, Dawn Taylor has endeared herself to many of us with her honest, articulate insights into what it has been like growing up with Ryley, who is now 8 years old. The following are excerpts from a recent letter I received from Dawn.

Learning to Run the Race

"I had hoped to write to you from Christmas Island, as I imagined I would write a more objective or positive letter rather than a predominantly negative one if I had corresponded earlier. I found though that when I thought of Ryley then I still harboured negative thoughts, and I needed more time to balance or reflect on life and put things in perspective.

"I know I am not alone in this train of events. It is a familiar dread in everyone's lives—the build-up to a summer break. It's almost like a marathon that you only just finish, and every year the race gets longer and the conditions more difficult. I guess it's when you finish the race and prepare for the next one that you look at how you ran the last one and make changes to your race plan.

"One of my changes is to be fitter physically, which will automatically help the mental aspect. I do resent not having enough time in my day for quiet reflection. I seem to be much too busy doing! So number two is time to reflect. Third is more time for Roland and I to spend together for doing things (don't smirk). We've given so much to the children over the last couple of years, and a short holiday or weekend away for just the two of us sounds extremely inviting to me at the moment. So there are my plans for the following year—how to go about achieving them is another thing altogether."

Behavior Problems—Handling Embarrassment with Style

"Ryley gets very frustrated and usually throws mega-tantrums which result in heavy destruction to his room. He needs to be actively involved in something with supervision and we have exhausted most avenues (e.g., pets—killed them; games—breaks them in a rage; friends—scares them with tantrums; computer—broke; TV—lasts a short while).

"Out-of-the-house tantrums, however, are usually more theatrical and appear when he feels out of control—*or* perhaps he wants to control. They usually involve an audience, where Ryley can inflict severe embarrassment through his varied and colorful (!) language and animal noises to achieve maximum attention. He also throws himself prostrate on the floor kicking madly at anyone who attempts to touch him. A pathetic sight. I laughed once because it really was so pathetic and was rewarded by shrivelling, poisonous looks from our audience. Why is a mother always at fault?

"Other times I watch him whilst trying to overcome my embarrassment, and I see fear in his face and eyes. Something in his mind just shuts down and he goes into overdrive 'losing it.' I suppose the easy answer would be to empower him with an 'out' for these circumstances when he's in genuine fear and out of control. An avenue of escape to divert his anger/fear should put him back in control, but what!? This needs thinking outside the square to a solution—perhaps you have some ideas or thoughts. *[Comments from readers?—Janalee]* I usually abandon whatever I was about to do and drag him out. I'm sure if I smacked him half the audience would applaud and the other half boo. Sometimes I think Ryley should get an academy award for his act. Once we're outside or in the car, he invariably makes wild promises of, 'I sorry—I never do that again. I love you, Mommy.' Reminds me so much of Matt's tantrums and how he wrote about the thing in his head that makes him do it. I also wonder if it is a lack of language or form of communication that forces them to resort to such violent means of attention or is it a vengeful act of power grabbing that drives them? And most of all, in all of this, where did I go wrong? There are always unanswered questions."

Siblings—They're Important, Too

"Has Sarah ever thought of participating in a follow-up to 'Sometimes I'm Mad . . . Sometimes I'm Glad'? I used to worry so much that Ryley would 'push' Avevska and Tris out of the house when they got older. I guess that's why we have spent so much time with them to help them feel important and loved and that their problems are just as important to us as Ryley's. Open communication, of course, plays a big part together with honesty. That's the hard part—to admit to your children that you're not superwoman and don't have an answer to everything. It's hard to discuss with one child the negative feelings you may have for the other. It's hard also to hear them pour out all their negative and hurtful feelings and thoughts about their brother and not to criticize them for rejecting him at times."

Respite—'You Deserve a Break Today!'

"It's also hard to keep adjusting life as a family with Ryley and then life as our family in respite. It's like we're two different families. Still, the more respite we get, the more loving and patient we all are with Ryley when he returns. We are adjusting, and these days I miss him when he's gone and I don't feel great despair of, 'Is this what it's like to be normal!?' I don't have to deal with all that guilt either, so all in all—I've come a long way and I've done well!! We have even reached the stage where I take Ryley out for breaks with just the two of us. Sometimes he's so cute and friendly and he tries so hard that my heart swells with pride for him. So now when there are really bad times I know they will pass, and we will survive, and we will be rewarded with the good times.

"I really believe this is all due to respite, and I feel very sorry for any family that is still groping around in the dark on their own, as we were. I am grateful to the institutions that now acknowledge the need for respite and for the volunteers who give their love and support. I am also grateful to all the parents before us who have 'fought' for this privilege that we now enjoy."

When you run the Prader-Willi marathon, the race doesn't get easier, you just get more skilled at running it, and hopefully you learn you don't have to run the race alone.

As Dawn so beautifully expressed, in order to run the race well, the torch should be passed from parent to parent and from parent to professional—and from the USA to Australia and back.

Board of Directors

PWSA Business Plan

Special Teams Created

by James G. Kane, PWSA Treasurer

In the March-April issue of The Gathered View, PWSA Board Chairman Curt Shacklett provided an overview of PWSA's evolving business plan, as developed during the January Board meeting in Atlanta. In this follow-up article, Jim Kane, chairman of the Board's Long-Range Planning Committee, describes in greater detail several of the new initiatives that have been launched.

In January, the Board of Directors of PWSA identified the four "Essential Functional Areas" of our Association. These are: (1) communications; (2) clearinghouse and funding source for research and scientific data; (3) state chapter support, development, and communication; and (4) organization and support for a national conference. There were other areas identified as being important; however, the Essential Functional Areas were judged to be the most critical to the Association's mission.

For certain officers and standing committees of the Board, specific "action steps" were identified and assigned. Each action step was given a certain priority and assigned resources, both in time and funding. An example is the Publication Committee's review of all existing PWSA products—both publications and audiovisual materials—which was given a high priority by Board members.

In addition, special teams were organized to work in several new areas. These teams are not necessarily standing committees and may be dissolved once they complete their tasks. Following is a description of four special teams and their assignments:

● **The PR Team** will develop a comprehensive public relations plan for the Association. In the past, PWSA primarily focused on serving the basic needs of families and state chapters, with less attention devoted to the public image or promotion of the organization. Given the growing competition for charitable contributions and funding and the increasing sophistication of the public relations efforts of other nonprofits, it is clear that PWSA needs to focus energy in this area.

● **The Liaison Team** will explore the possibility of establishing relationships with companies and organizations that have areas of common interest with PWSA, including drug manufacturers, food and dietary supplement producers, universities, and major foundations. Both parties could benefit from sharing ideas, information, and current research in the common interest area. Groups that could provide funding for our Association would be key targets. They might be asked to fund a particular project or action step in PWSA's business plan or simply to provide ongoing annual support of our Association's work. We do not have to be in this alone! But we need to reach out in order to garner support from as many new sources as we can.

● **The Organization Team** is evaluating ways the Association can cooperate with and learn from organizations similar to our own. PWS is considered an orphan syndrome, based on its relatively rare occurrence. There are innumerable syndromes and diseases with similarly low incidence rates, and many are represented by associations. The Organization Team is studying ways of increasing our contact and cooperation with those sister groups. Experience is valuable, and sharing ours can benefit PWSA.

● **The Data Team** is analyzing the use of computers in the National Office. The world of office automation is moving extraordinarily fast, and the latest and best methods are difficult to harness. The National Office currently uses several computers of varying ages and capabilities. This team will evaluate the best way to employ the computers and assess the need for updated equipment. Our staff and volunteers can probably all work better, faster, and more enjoyably as a result!

These four teams are working away and will report to the Board of Directors in July.

If you have a particular sl resource, or contact to help with the work of any of these teams, please call the National Office (or return the form on page 11 of this issue) and VOLUNTEER!

Position Available:

Secretary for the Board of Directors of PWSA (USA)

Responsibilities:

"The secretary shall have the responsibility to act as a Recording Secretary for the transactions and procedures of the Board at all Board meetings." -- *PWSA bylaws*

There are two Board of Directors meetings each year--one held at the national conference and the other in January--at various locations throughout the country. You would be required to attend both meetings, at your own expense.

The secretary is appointed by the Board for a one-year term, with no limit on the number of terms.

The position is available beginning with the upcoming Board meeting in Scottsdale, Arizona, July 14, 1993.

Qualifications:

You must be able to take accurate notes quickly. Any PWSA member who is interested should call the National Office or return the volunteer form included in this issue of *The Gathered View* as soon as possible.

Compensation:

Expenses for officers are reimbursed up to $500 per year. You also receive tremendous support and a sense of participation in the Association like you have never felt before.

Board Election Details

The Prader-Willi Syndrome Association (USA) is headed by a 12-member Board of Directors. Each year at the national conference the general membership elects a certain number of Directors to serve a three-year term.

This year there are two Board positions to be filled. Directors completing their terms are Suzanne B. Cassidy, M.D., who is running for re-election, and Curt Shacklett, current Chairperson of the Board. Mr. Shacklett has chosen to step off the Board in order to dedicate more time to obtaining services in Oklahoma. He will be greatly missed.

At the general membership meeting at the conference, the floor will be open for nominations for the other position.

Each individual or organization paid membership is entitled to one vote. Each family paid membership is entitled to two votes (which cannot be voted by one person). You may cast one vote for each Board vacancy, but you may not cast more than one vote for each candidate.

Members who are unable to attend are urged to cast a proxy ballot. "Proxy" means you are designating someone who will be at the conference to represent you because you cannot be present to vote yourself. This could be any member of PWSA or of the Board of Directors. You will find the proxy ballot on page 12. Participate in your Association by voting!!

Suzanne B. Cassidy, M.D.

Susie, as she is known by many, joined PWSA in 1983 and became a member of the Board of Directors in 1985. She is also a member of the PWSA Scientific Advisory Board. A pediatrician and geneticist, she first became interested in PWS in 1980 during her genetics fellowship at the U. of Washington. PWS has been her major area of research since her first project to look at the correlation between the chromosome deletion and clinical findings in this condition.

She has been an Associate Professor of Pediatrics at the U. of Arizona College of Medicine since 1988. Prior to that, she held the same position at the U. of Connecticut, and at both places she headed PW clinics. She has written and spoken extensively on the syndrome on a professional level, but she is also known for her unique ability to communicate difficult and highly technical concepts in clear terms that readers of *The Gathered View* and audiences at PWS conferences can understand.

In 1991, she organized an international scientific workshop on PWS which was held in The Netherlands; a book on the proceedings of that meeting has recently been published.

This year marks the end of Susie's third term on the Board, and we are pleased to announce her willingness to run for a fourth term.

Conference Notes

* Reminder to all who have registered for the conference: make your hotel reservations as well.
* Registered participants of the Youth/Adult Activity Program will begin their days of fun with a pool olympic event. Please come dressed in swim suits with a change of clothing on Thursday morning.

Spotlight

A Proud Eagle

Jim and Carol Draffen of Memphis, Tennessee, sent PWSA an invitation to their son Stephen's Eagle Scout Court of Honor ceremony. They enclosed the following note:

"We want to inform PWSA of Stephen's Eagle Ceremony [held on April 4, 1993]. To our knowledge, he is the first person with PWS to receive Eagle in the Boy Scouts of America.

"He is attending Woodale High School in Memphis, where he is a sophomore. He is 18 and very active in school activities."

We're delighted to share the good news that several other young men with PWS have also achieved this high honor.

Our congratulations to Stephen !

The New World of Genetics

From Gregor Mendel, the Father of Genetics, to Genomic Imprinting of Genes on Chromosome 15: We've Come a Long Way

by Maria J. Mascari, Ph.D., Division of Genetics, Milton S. Hershey Medical Center

It took 25 years from the initial description of Prader-Willi syndrome (PWS) in 1956 till researchers discovered that a majority of affected individuals were missing a portion of chromosome 15 (Ledbetter et al. 1981, 1982). This discovery, along with improved clinical awareness of the disorder, enabled many families to put an end to the long list of unanswered questions. Despite these advances, however, there remained 30 to 40 percent of individuals with PWS who did not have a deletion of chromosome 15; so, many parents still had no explanation for their child's diagnosis.

Maternal Disomy Discovered

The discovery of "maternal disomy" in two individuals with Prader-Willi syndrome (Nicholls et al. 1989) represented a major breakthrough in understanding the etiology [cause] of PWS. "Maternal disomy" refers to the situation where an individual inherits both chromosomes of a pair (the #15s, for example) from the mother and does not appear to inherit the respective chromosome from the father. Typically, we inherit one chromosome of the pair from each parent.

This discovery prompted two subsequent studies which showed that approximately 20 to 25 percent of the PWS individuals exhibit maternal disomy for chromosome 15 (Robinson et al. 1991; Mascari et al. 1992). Thus, molecular diagnosis of PWS, looking for either a deletion or maternal disomy, was now possible in up to 90 to 95 percent of affected individuals. Consequently, only 5 to 10 percent of PWS individuals continue to demonstrate normal inheritance patterns with the current level of molecular technology.

Along with improvements in both clinical and molecular diagnosis came a number of questions generated by the new discoveries. Butler and Palmer (1983) had previously reported on a small group of PWS individuals with known cytogenetic deletions in which the chromosome carrying the deletion was consistently of paternal origin. Their results, when analyzed statistically, were unlikely to have occurred by chance alone and were subsequently confirmed by molecular studies. It appears that, although both of the paternal chromosome 15s are normal, a sporadic event occurs at some point during the formation of the sperm such that one of the chromosome 15s is missing a portion of genetic material. ***It is important to emphasize that this type of event occurs sporadically in the general population and is one over which we have no control.***

Although this discovery was intriguing from a scientific point of view, the question still remained as to its potential significance to the etiology of Prader-Willi syndrome. Once maternal disomy was described in PWS, the pieces of the puzzle began to fit into place. One might initially question why an individual with maternal disomy for chromosome 15 would have PWS since technically they have the correct amount of genetic information, but instead of inheriting one chromosome 15 from each parent as Mendel's laws of genetics would dictate, they inherit both chromosomes of the pair from their mother. One might also question why two different molecular abnormalities (paternal deletion and maternal disomy) can both cause PWS, even though they originate via different mechanisms.

Genomic Imprinting Explains Mysteries

The answers to these questions are best explained by a new genetic concept which suggests that some genes are expressed differently depending on from which parent supplies them. This concept, referred to as the theory of "genomic imprinting," implies that some chromosomes or regions of chromosomes within the egg and sperm may be marked differently, i.e., maternal vs. paternal. Although two completely different mechanisms are involved in the etiology of PWS (deletion vs. disomy), both share in common the absence of genetic information normally supplied from the father. Evidently, there is one or more genes on chromosome 15 which must come from the father in order for normal development to occur. The reception of the corresponding gene or genes from the mother, as in maternal disomy, cannot compensate for the loss of paternal information because the maternal gene(s) are inactivated and thus not capable of being expressed. Likewise, there are some genes which must come from the mother in order for normal development to occur and for which the father's gene(s) cannot compensate, as demonstrated in Angelman syndrome.

An additional observation made possible by the detection of maternal disomy in PWS was its association with advanced maternal age. In other words, the mothers of PWS children/adults with maternal disomy tend to be older than the mothers of those who have a paternal deletion. This observation is not a complete surprise since the occurrence of Down syndrome and some other types of chromosomal abnormalities have previously been associated with maternal age.

Disomy May Originate from Trisomy

In 1980, long before maternal disomy was even described in Prader-Willi syndrome, Engel suggested a number of possible mechanisms for the etiology of uniparental (either maternal or paternal) disomy. One mechanism in particular suggested that maternal disomy could result from the fertilization of an egg containing two copies of a chromosome (#15, for example) by a sperm carrying the expected one copy of that chromosome. The fertilized egg resulting from that union would then carry three copies of the involved chromosome, a situation referred to as "trisomy," which is usually not compatible with life. Subsequent loss of the paternal chromosome in this example would then result in maternal disomy for chromosome 15, a situation which, although compatible with life, would result in a child with Prader-Willi syndrome.

This mechanism was further supported by two independent case reports

(Cassidy et al. 1992, Purvis-Smith et al. 1992) which each described the prenatal diagnosis of a fetus with Trisomy 15 via chorionic villus sampling at 9 to 11 weeks gestation, followed by the identification of a normal chromosomal complement at amniocentesis, suggesting loss of the extra chromosome. In each case, the baby was hypotonic at birth, exhibited failure to thrive, and was subsequently diagnosed to have Prader-Willi syndrome. Theoretically, the loss of the extra chromosome represented a selection for survival on the part of the fetus (Rogan et al. 1991), since Trisomy 15 is not compatible with life.

Diagnostic Testing for Deletions, Disomy

As a direct result of the intense research efforts over the past 10 to 15 years, a variety of diagnostic tests are capable of confirming a clinical diagnosis or suspicion of Prader-Willi syndrome. High resolution chromosomal studies, where the chromosomes are stretched as long as possible, remains the first and foremost method of laboratory diagnosis. Although the high resolution chromosomal studies, sometimes referred to as prometaphase studies, are capable of detecting many of the PWS individuals with deletions, they cannot detect small deletions that are not visible under the microscope, nor can they detect maternal disomy. Likewise, a newer laboratory method, called "fluorescent in situ hybridization" (or FISH, for short) can detect deletions but not maternal disomy. Currently, molecular studies are the only method to detect both deletions and maternal disomy in PWS. Typically, these studies involve obtaining blood samples not only from the affected individual but also from the biological parents to follow the reception of the child's chromosome 15s from the parents. Recently, Driscoll et al. (1992) described a new and innovative method of molecular diagnosis which can detect alterations in the methylation of a person's genetic material. This approach can detect methylation changes in PWS individuals with either a paternal deletion or maternal disomy and has the advantage that both parents do not have to be available for study.

The research on Prader-Willi syndrome over the past 10 to 15 years has not only improved our diagnostic capabilities but has challenged us to reevaluate our understanding of genetic inheritance. In addition, these discoveries are helping to unravel the mysteries of the inheritance of other genetic disorders, including a number of syndromes that predispose toward cancer formation.

In regards to diagnostic testing for Prader-Willi syndrome, high resolution chromosomal studies are widely available on a clinical basis throughout the United States. In contrast, molecular testing is only available at a few centers and on an investigational basis.

Questions regarding molecular testing can be directed to:

Maria J. Mascari, Ph.D., or
Peter K. Rogan, Ph.D.
Division of Genetics
Department of Pediatrics
Milton S. Hershey Medical Center
500 University Drive
Hershey, Pennsylvania 17033
Tel.: (717) 531-8414
Fax: (717) 531-8985

Nutrition in Group Homes: A Research Summary

by Peggy Pipes, R.D., M.P.H., Nutritionist, Child Development Mental Retardation Center, University of Washington, and PWSA Scientific Advisory Board

A questionnaire survey to determine anthropometric data collected and diet-related problems was conducted of 25 group homes that house residents with the Prader-Willi syndrome. Eighteen of the 25 questionnaires were returned.

Survey Highlights

◆ The mean age of residents was 25 plus or minus 8.4 years. Sixteen of the homes reported that they locked their kitchens at night; 12 found stealing and hoarding of food to be a problem.

◆ Fourteen of the homes responded that they did not take skinfold measurements, 15 that they did not take arm circumference. Only two estimated body fat. Almost half of the homes weighed the residents weekly. A registered dietitian determined ideal weight in eight homes.

◆ One-third of residents had been successful in weight loss and were on maintenance diets; the remainder were on weight reduction diets. Residents consumed a mean of 1,000-1,500 kcal/day.

◆ When queried about the greatest challenge, respondents mentioned menu planning and determining an appropriate weight for each client. Another challenge of note was maintenance of desired weight when clients are on leave from the facilities.

continued on page 8

New PW Clinic in Pa.

A specialized clinic has been established at Hershey Medical Center in Hershey, Pennsylvania, to provide a comprehensive approach to the specific needs of persons with Prader-Willi syndrome, as well as to respond to the concerns and needs of their parents. Opened in Nov. 1992, the clinic now sees two to four families each month.

The Prader-Willi syndrome clinic team is headed by Dr. Roger Ladda, Margie Morris, R.D., and Maria J. Mascari, Ph.D., with referrals to other team specialists made on an individualized basis. Each visit includes a medical examination, physical growth assessment, and review of developmental/educational progress. In addition, both nutritional and social services are available to address specific concerns. For information contact Maria Mascari, Ph.D., Genetic Counselor, at 717-531-8414. *Editors' note: We invite Prader-Willi syndrome clinics across the country to write about their services and staff for publication in The Gathered View.*

continued from page 7

Recommendations

Clearly, guidelines are needed to help practitioners develop strategies for weight management in group homes. Recommendations were made as follows:

- Determine ideal weight; use of body mass index was suggested. A nomogram is included in the article.
  [Editor's note: This is a chart that can be used to compute body mass index for persons with PWS.]
  A supplemental method that should be considered is circumference measurements.
- Provide medical monitoring.
- Include an exercise component in daily activities.
- Develop an appropriate nutrition component. Many homes utilize a basic 1,000 kcal/day diet, then increase or decrease dietary energy as individually indicated. Appealing meals should be planned; individual food preferences and special occasions should be respected. A nutrition education program for the residents should be part of the plan.
- Behavior modification should be part of the plan. An interdisciplinary approach is needed.
- The program should be carefully monitored.

Reference: Hoffman, C.J., Aultman, D., and Pipes, P.: A nutrition survey of and recommendations for individuals with Prader-Willi syndrome who live in group homes, *Journal of the American Dietetic Association*, July 1992, 92:823-830, 833.

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Memorials:
(*In Memory of* - Contributor)

Peggy Ashbach - Marilee Kirk, Gordon Boen, Don & Beverly Simmelink, Dennis & Gail Kelce, First National Bank River Falls, Claudia Johnson, James & Patrice Pederson, Ed & Sandra List, North Star Central Morgans, Jeanne & David Ahrenholz, Shirley Orlando, Mr. & Mrs. Rueben Kaplan, Sally & Eves Whitney, Jacquelyn & Lee Millet, Kennametal Inc.

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Shaunna Sweeney - United Strathmore Employees Fund

Dr. Harold Canter - Ida & Judy Singer

Shirley Jurgenson - Marilyn Battersby

Chet Erickson - Marilyn & Duane Sucha

Ms.Aniela Majka - Bill & Erna Anderson

Frank Love - Joan & Tom Hughes-Peifer

Ask the Professionals

Q: ***Is it true that chewing gum after meals helps to prevent tooth decay? If so, should it be any special kind of gum, and how does it work?*** [Submitted by a young person with PWS, Julie Mitchell, 23, from Pittsburgh, Pa.]

Providing the answer is Barry Margolis, a general dentist and a partner in a group dental practice in West Newton, Massachusetts. Dr. Margolis served in the National Health Service Corps prior to entering private practice and is the father of a 6-year-old daughter with PWS.

A: These are excellent questions with important consequences for people with PWS. Yes, it is true that gum chewing after meals helps to prevent tooth decay. Chewing either sugar-sweetened (sucrose) or sugarless gum after eating reduces cavity formation and can be recommended as an adjunct to normal oral hygience when brushing and flossing are not possible. However, chewing sugarless gum is a more effective way of reducing cavities when compared with chewing sugar-containing gum.

The brand of sugarless gum is less important. Dental studies indicate that sugarless gums (sweetened with xylitol or sorbitol) are all nearly equally effective in reducing cavities. A check of the ingredients on the label will tell you what type of sweetener is used in a particular gum.

Before explaining how this works, let's briefly examine how cavities are formed.

The following formulas illustrate the process:

Sugar + Bacteria = Acid Production
Acid + Teeth = Cavities

Therefore, two things are needed to form cavities: sugar and bacteria. No matter how well one brushes and flosses, there will always be bacteria present in the mouth. Elimination of bacteria is beyond our control.

However, sugar consumption is something that we can control. If you must eat sugar (and there are forms of sugar in many foods we eat), the dental consequence is that your saliva will become acidic within five minutes and your teeth will continue to be bathed in this acidic saliva for 30 to 105 minutes.

The good news is that chewing gum after eating sugar stimulates increased salivary flow, which dilutes and neutralizes the acid, thereby reducing or eliminating the potential for tooth decay.

In addition, the increased salivary flow initiated by chewing gum supplies more calcium to the teeth, which helps to harden and remineralize their enamel. Therefore, not only does chewing gum help to prevent new decay, but it potentially can help to remineralize small cavities that have already started to form!

It is important, however, to realize that the studies that were done to test the effects of gum-chewing on salivary flow and decay rate were not done using people who have PWS. This may represent a significant distinction, because individuals with PWS generally have less salivary flow (thicker and stickier saliva) than that of the general population. This could result in less of a neutralizing effect than seen in the research. However, it is still likely that some degree of benefit would still be obtained from chewing gum. More research with the PWS population is needed to substantiate these conclusions.

Recommendations:

- Gum-chewing should begin as soon after eating a meal as possible and be continued for at least 20 to 30 minutes.
- Sticky, retentive foods should be avoided whenever possible (raisins, Milky Way bars, etc.). Dental studies clearly show that gum-chewing for 30 minutes will not completely remove these types of food from the teeth. Therefore, once the gum-chewing is stopped, the sugar that remains on the teeth will result in new acid formation and the potential for cavities. On the other hand, sugars consumed in liquid form, such as juices, are washed away almost immediately and completely by chewing gum and are therefore more desirable.
- Frequency of sugar intake (and snacking in general) should be kept to a minimum, because every time sugar is consumed it produces acid for 20 minutes.
- Brushing and flossing are still the best methods of removing food debris and plaque from the teeth. Remember the formulas that result in cavities. If the sugar is brushed off the tooth surfaces, acid will not be produced directly on the teeth.

Questions, questions, questions. . .

Got any you'd like to see answered in *The Gathered View*? We'd be glad to pose your questions either to other parents or to professionals in any field, but we need to know what your questions and concerns are!
Let us know by calling, writing, or faxing to the National Office anytime.

The Gathered View Question Box,
PWSA (USA), 1821 University Ave. W., #N356, St. Paul, MN 55104
Telephone: 800-926-4797 or locally 612-641-1955, fax: 612-641-1952.

From the Home Front

The Founding Family

Our son, Curtis, who was diagnosed with Prader-Willi syndrome in 1971 as an infant, was the impetus for our starting to work in 1974 on founding the Prader-Willi Syndrome Association. We thought you might be interested in reading what he has written about himself. He decided to write this letter after reading the article in the November-December 1992 issue of *The Gathered View* written by another young man with PWS ["Will the Real Merrill Please Stand Up?"].

Gene and Fausta Deterling
Orono, Minnesota

Putting Others' Hunger First

My 9-year-old grandson Karl was diagnosed as having PWS soon after birth, even though he had no apparent 15th chromosome abnormality. He has always been on a strict diet, which he accepts as normal; but, as with others with PWS, meal times are the most important times of the day.

For the past three years, Karl has participated in the annual 40-hour famine with his Sunday school. The first year he did a 20-hour stint, which was usual for the younger children, but for the last two years has insisted on fasting for the 40 hours. He has succeeded each time, raising each year $200 for the world's starving.

Karl has always been very concerned for the people of Third World countries and can understand better than most, I suppose, what it feels like to be continually hungry.

We're all very proud of him as it is a great achievement for any 9-year-old, let alone one with PWS.

Lesley Greenall
Woodbourne, New Zealand

(We're proud of you, too, Karl -- Congratulations on a tremendous accomplishment!)

Dear Association Members:

My name is CURTIS CHARLES DETERLING, and this is my story about my life at present. I live in a group home for people with all different disabilities and mental retardation problems. I live in Baker House which is a living unit arrangement place. I live with 12 other residents of different ages. There are 8 PRADER-WILLIES in the house. There is one hearing disabled resident with DOWN SYNDROME. She can hear partially out of both ears. Her name is LANNIE. The other residents are MICHAEL, ROBERT, MICHAEL, THERESA, KAREN, VIRGINIA AND KARI. Those are people like me who have the same syndrome as me. The other residents are RONALD, TIMOTHY, JEFFREY, and RITA.

I am 22 years old and have lived at this home for almost 6 years. It is called LAURA BAKER SCHOOL. It is located in NORTHFIELD, MINNESOTA. It is the place where the great all-American bank raid took place. The one that was done by JESSIE JAMES and the YOUNGER BROTHERS.

This group home is in RICE COUNTY, MN. I go to work at EPIC CENTER, and then some days I go to THE SUPER 8 MOTEL in DUNDAS, MINNESOTA. I like where I live because they give us good care there, which is especially DIETING NUTRITION.

My favorite hobbies are basketball, football, baseball, softball, hockey, ice skating, crosscountry skiing, swimming, seashell collecting, weightlifting, bodybuilding, art, dancing, listening to music, walking, running, jogging, jumping, skipping, galloping, hopping, rolling, bikeriding, shoveling snow, chopping ice, sightseeing, vacationing to different places, going to the beach by the ocean in CAPE COD, MASSACHUSETTS, saltwater wave jumping, reading, activity books, puzzlemaking, jumproping, frizzbie throwing, nerf boomerang throwing, supersoaker skwirt gun fights, pogo ball jumping, taking photos, being a cowboy, collecting caps, cooking, baking, the medical field and animal loving, and also taking care of my niece. My interests are the medical field, getting my driver's license, getting married, getting a house of my own and having a family of my own. I like being with my family at home for holidays, except for Halloween.

Curtis Charles Deterling
Northfield, Minnesota

(Thanks for taking time from all those activities to write to us, Curtis! And we compliment you on your excellent writing and spelling.)

Mississippi Group Home Has Openings

A new six-person residential facility, designed specifically for individuals with PWS, has recently opened in Mississippi with two residents. Operated by the North Mississippi Regional Center, "Fernwood" is located at Two Industrial Drive, Highway 7 South, in Oxford.

For information about placement in this facility, please contact Dr. Carole Haney,PO Box 967 Hwy . 7 South, Oxford, MS, 38655, or phone (601) 234-1476.

Call for Volunteers—You are Needed!

by Tere Schaefer

One of the discussions the Board of Directors had during its mid-winter planning session was about the importance of volunteers to an organization such as ours--the volunteers we already have and the new ones we will need to accomplish our goals.

Each one of us volunteers for something at some time in our lives, and usually it is when someone calls directly and asks. Since this newsletter is very much our voice to the members, pretend the phone just rang, and I am asking you to volunteer. There's quite a choice of things you could do:

* Send the National Office staff chocolates. (Just kidding!)
* Generate mailing and phone lists of the resources (doctors, dietitians, other specialists and service providers, agencies, and organizations) in your state for other parents and professionals.
* Make contacts with companies in your state to request funding grants for PWSA projects.
* Organize a local fundraiser. (A member in Ohio raised more than $5,000 by organizing a dance!)
* Participate in PWS Awareness Day (details to be outlined in the future)
* Write articles or responses to questions that appear in *The Gathered View.*
* Assist the Publications Committee in its review and revision of PWSA publications.
* Serve on a committee of the Board (listed below).
* Serve as Secretary for the Board of Directors (see job description on page 4).

We are serious about these requests! There's a great deal of work to be done, and we need both large and small commitments of time and expertise from the membership. Return the form below if you are ready to help.
Finally, if you live near the National Office, stop in any day, and we can put you to work for the benefit of all.

I Want to Get Involved in PWSA

Please contact me about ...

❒ **Serving on a committee of the PWSA Board**

- ❒ Board Nominations
- ❒ Investment
- ❒ Bylaws Review
- ❒ Long-range Planning
- ❒ Crisis Intervention and Training (CIT)
- ❒ National Conference
- ❒ Publications/Education
- ❒ Fund Raising

❒ **Assisting one of the PWSA Board's special teams**

(See article on page 4 of this issue for descriptions.)

- ❒ Public Relations Team
- ❒ Liaison Team
- ❒ Organization Team
- ❒ Data Team

❒ **Assisting on specific PWSA publication/video projects**

(Check your areas of expertise)

- ❒ Writing/editing
- ❒ Word processing
- ❒ Printing/publishing
- ❒ Graphics/art
- ❒ Publication layout/design
- ❒ Video production
- ❒ Other:_____

❒ **Serving as Secretary to the PWSA Board of Directors** (See job description, page 4.)

Name:_____

Telephone: _____days _____evenings _____weekends

PWSA (USA) PROXY BALLOT

(Family Memberships are entitled to two votes)

I(we) , _____, appoint the following person to vote as my (our) proxy in all voting transpiring during the 1993 general meeting. [Includes voting for board of directors and any issues which may come before the board at this meeting.]

Proxy Name: _____

Please vote on my behalf following these guidelines:

Board of Directors:

❒ Suzanne B. Cassidy, M.D.

❒ _____

write-in(s):

This column for second family vote only

❒ S. B. Cassidy, M.D.

❒ _____

write-in(s):

Signature (s)/Date: _____ _____

_____ _____

Proxy must be returned to National Office by Friday, July 2, 1993.

This is your official ballot. No reproductions of this form or phone calls will be acceptable.

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 publication is prohibited. Quotations may be used upon credit given to PWSA(USA). **The Association gratefully acknowledges its volunteer editors, Lota Mitchell and Linda Keder.** If you are interested in a membership, dues are $21 for an individual, $26 per family, $31 per agency/ professional. Send dues, change of address, or letters to: 1821 University Ave. W., Suite N356, St.Paul, MN 55104-2801. Questions or comments regarding this publication or PWS call: 800-926-4797 or 612-641-1955 or Fax 612-641-1952.

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