



The *Gathered View*

December 1995
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National Newsletter of the Prader-Willi Syndrome Association (USA)

President's Message

*Oklahoma City
November 17, 1995*

If ever a sequence of events can change the fabric of our lives, it happened at 9:02 a.m. on April 19th in Oklahoma City. The emotions that have surfaced since the tragic bombing—grief, empathy, compassion, and persistence—we as parents, siblings, or caregivers of children with Prader-Willi syndrome have also faced daily and felt deeply. From that moment in our lives when we “knew something was wrong” or received the correct diagnosis to a most recent behavioral outburst, the impact on our families is real and often painful. We all have incredible courage to tackle, on a daily basis, the highs and lows of this syndrome that becomes woven into every aspect of our lives. We constantly see a better life for those people with Prader-Willi syndrome.

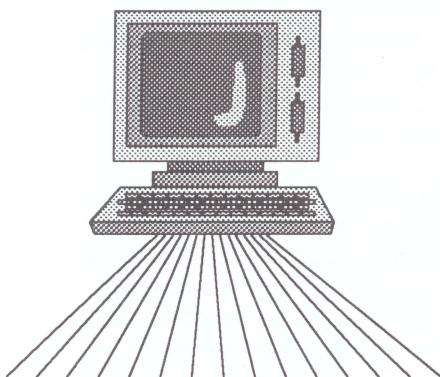
One of the many goals of the PWSA (USA) is to provide the support for families through information and education about the syndrome. As we network with each other about our current situations, we are always learning new ways to help us cope.

As we saw this year in Oklahoma City, there can be great strength in a community when it is forced to act under adverse conditions. Many positive things happened because the families bonded together to see their situation through. Though the passage of time affected people in different ways, it was “the event” that caused the change in their lives. The families and many dedicated professionals of our Association are what has created “our community” and who help us deal with the events in our lives. It is our hope that through continued support and dedication our “community” will continue to be strong.

May we take this opportunity to wish friends and families of Prader-Willi worldwide a very joyous Holiday Season. We wish in the New Year as one of hope for our children with Prader-Willi syndrome.

Sincerely,

*Jerry R. Park
President, PWSA (USA)*



Announcing ...

The PWSA (USA) World Wide Web Page

*on the Internet for information
about Prader-Willi syndrome and
PWSA (USA)*

See page 3 for details

You can now contact PWSA via electronic mail (e-mail)!

America Online users can send messages to us at: **pwsa usa**

E-mail from other commercial services or the Internet should be addressed to:

pwsa usa@aol.com

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The Prader-Willi Syndrome Association (USA)

2510 S. Brentwood Blvd., Suite 220
St. Louis, MO 63144-2326

(800) 926-4797 or (314) 962-7644

8:00 a.m.-4:00 p.m. CDLST

Fax (314) 962-7869

E-mail: pwsa usa@aol.com

National Office Staff

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Office Manager - Maureen A. Brown

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Out of The Office

by: Russ Myler, Executive Director

While the news I want to tell you is very positive for Prader-Willi syndrome research, this is a very difficult letter for me to write just as it will be a very difficult one for you to read. It concerns loss of a loved one and decisions necessary from that

loss. I hope you will understand the importance of this letter from me to you.

For many years researchers have told us about the need for studies of brain tissue to further their understanding of the physical consequences of Prader-Willi syndrome. Genetic research and clinical treatment can infer what has happened, but only tissue studies can confirm what are now educated guesses. Such studies can further the ability to treat the effects of the syndrome and may bring us closer to understanding the cause.

Recognizing this need, the PWSA (USA) Board of Directors asked the Scientific Advisory Board to pursue this issue. Drs. Suzanne Cassidy, David Ledbetter and Merlin Butler are acting as our medical advisors on this very important question. Over the past six months they have established a process whereby parents, professionals, caretakers and others may get information, get questions answered and make arrangements for donations.

I know this decision is hard to make, having to make it twice myself. But I also know that these decisions will make a brighter future for those who benefit from the results of these donations.

For information please contact:

Suzanne B. Cassidy, M.D. 216-844-3936, David Ledbetter, Ph.D. 301-402-2011, or Merlin Butler, M.D. 615-322-7601

IN MEMORIAM

The sympathy of the Prader-Willi Syndrome Association is extended to Executive Director Russ Myler in the loss of his mother, Lorene Miler. Mrs. Miler passed away on October 9 after a long and difficult illness.

The Gathered View (ISSN 1077-9965) is published bimonthly by the Prader-Willi Syndrome Association (USA) as a benefit of membership. Annual membership dues are: \$30 Individual, \$35 Family, and \$40 Agencies/Professionals for U.S. members and \$40, \$45, and \$50 (US Funds), respectively, for members outside the United States.

Opinions expressed in *The Gathered View* are those of the authors or editors and do not necessarily reflect the views of the officers and board of directors of PWSA (USA). *The Gathered View* welcomes articles, letters, personal stories and photographs, and news of interest to those concerned with Prader-Willi syndrome.

Editors: Linda Keder, Silver Spring, Md.
Lota Mitchell, M.S.W., Pittsburgh, Pa.

Communications regarding *The Gathered View* or PWSA membership should be directed to the national office of PWSA (USA), 2510 S. Brentwood Boulevard, Suite 220, St. Louis, MO 63144-2326. Telephone 1-800-926-4797, or (314) 962-7644 in the St. Louis area. Fax (314) 962-7869. E-mail: pwsa usa@aol.com.

PWSA on the Information Highway

by Mike Keder, Chair, PWSA Data Team

PWSA has established a World Wide Web (WWW) page on the Internet. This will allow us to give and receive information about PWS and PWSA on one of the trendier communications media. The initial offering consists of the following sections, but as with most computer projects, it will grow over time.

- What Is Prader-Willi Syndrome?
- What Is PWSA?
- State Chapters and Affiliated Organizations
- Publications
- Frequently Asked Questions About Prader-Willi Syndrome

The choice of topics was based on the results of a survey sent to various groups within PWSA and the availability of information. Future topics under consideration include: resource listings by state (physicians, group homes, etc.), international contacts, research updates, expanded medical information, meetings/events, and listings of PWS-related articles. We are also working with others to develop links between their Web sites and ours.

Mike Larson, the Wisconsin chapter president, developed our Web page and is responsible for adding material as it is approved. Most of the credit for the Web page goes to him.

You may access the PWSA Web page directly from the Internet or through most of the commercial services (America Online, Compuserve, Prodigy) with a "Web browser" such as Spy Mosaic or Netscape.

We welcome feedback from all PWSA members and home page users. Send comments, questions, and suggestions to the Data Team in care of the PWSA national office. Try our new E-mail address: pwsa_usa@aol.com.

Map to the PWSA Home Page

To access the PWSA World Wide Web page, you will need the following address:

http://www.athenet.net/~pwsa_usa/index.html

(Note: It must be typed exactly as it appears above, with all symbols and punctuation.)

From the Internet — Enter the address in your browser. There should be a way to save it so you won't have to re-key it each time.

From America Online (AOL) Version 2.5 — From the main menu select INTERNET CONNECTION. On the next screen select the WWW icon. When the next screen appears, type the address into the address box at the top of the screen.

From Compuserve — You need Version 1.4 and the Spy Mosaic browser, which can be downloaded. Select the Spy Mosaic icon from the main group. Once you are connected, select the "Hot List" icon and add the address under an appropriate category. Choose Hot List from the menu bar and double click on the name you gave the address. After the initial setup, just repeat the last step after connecting.

INSURANCE ALERT

Managed care option may prohibit rehabilitative in-patient care for a family member with PWS

Today, many—or even most—of us are being asked to choose between traditional insurance coverage and managed care programs such as HMOs. It appears that HMOs, for the most part, do not seem to see the necessity for in-patient care of persons with PWS, particularly rehabilitative-type care for the problems of the syndrome such as behavior.

Choice of insurance is particularly pertinent when parents become older and become eligible for Medicare. When the parent of a person with PWS goes on Medicare, the child with PWS, no matter what age, will also become a Medicare recipient. Medicare recognizes the need for in-patient care for PWS, and it usually is in agreement with physicians recommending it. However, many Medicare recipients are being encouraged to choose the managed care program these days, which is very likely to result in the person with PWS only being eligible for services for severe medical illnesses (heart failure and pulmonary failure, for example) and not for rehabilitation or behavioral treatment.

Medicaid (which uses state funds for medical care for people with low income who qualify) in many states is run by managed care programs, so those recipients may have no choice. But Medicare recipients do have the option: regular insurance program or managed care program. Often, free medications or minimum charge for medications is offered as an incentive for people to sign up for managed care.

Of course, managed care is held up as one way to control medical costs, and it may indeed be helping to control costs in some situations. But it does limit the patient's options.

If you have a choice, investigate carefully before you make your decision.



We are excited to offer an adult evening trip to the St. Charles Casino which will coincide with the Youth and Adult Activity Program going on a trip to the Magic House. The Casino trip will be Thursday evening and is free (except for "spending" money, \$500 max.) but will require advance sign-up. Be on the lookout for more information when your registration packet arrives. You can make your travel arrangements and hotel reservations now by calling:



Pat Mann
Regency Travel
1-800-784-7581

Chapters

Welcome, Nebraska Chapter!

Our congratulations to families in Nebraska for becoming PWSA's newest state chapter. During last spring's Awareness Day activities, the Nebraska group—led by now Chapter President Roger Rhoads—raised one of the largest state contributions for PWSA through a Bowl-a-thon, signed proclamations with the Nebraska governor and the City of Omaha, and spread the word about PWS through the news media and various other outreach efforts.

Chapter Contact Changes

The last issue of *The Gathered View* listed chapter presidents and contact persons. We have updates or corrections for three states:

Arizona—Christy Montgomery (new contact person),
602-964-6322

Florida—Roda Guenther (new president),
941-542-4931

Missouri—Paula Kollarik, (corrected phone number)
314-931-0920

Scientific Advisory Board Changes

PWSA's Scientific Advisory Board is a selected group of individuals with expertise and demonstrated interest in research on PWS. Whenever possible, people who have documented their interest through attendance at PWSA annual meetings, including the Scientific Conference, and who are actively publishing on the condition are chosen. Generally, the chair of the SAB is chosen by the PWSA board of directors, and this individual chooses the members of the committee, pending approval by the board of directors. Its major functions are to review grants and make recommendations to the board of directors for funding; to encourage and promote research on PWS; to advise the board of directors with regard to medical and scientific issues; and to help answer questions from those requesting information and advice from PWSA.

Vanja Holm, M.D., a pediatric developmental specialist, was the first chair of the SAB, and her tenure lasted until 1994, when Suzanne Cassidy, M.D., a pediatrician and medical geneticist, took the position. Dr. Holm remains a member of the SAB. Recently, the board of directors decided that members of the SAB should have three-year terms on the committee, with the option for one consecutive renewal. In this way there is opportunity for new members, with presumably new ideas and approaches, to benefit PWSA. During this process of change, three long-standing members of the committee, who have made important past contributions, have chosen to resign.

Peggy Pipes, R.D., is a nutritionist who has worked with Dr. Holm's PWS clinic at the University of Washington for many years. She has written much and taught many young nutritionists about PWS. With her recent retirement, she has chosen to resign from the SAB. Her many years of contribution to PWSA will be long remembered.

Bryan Hall, M.D., a pediatrician and dysmorphologist, has also been a member of the SAB for many years. He has written extensively about PWS in the pediatric literature and has been a significant contributing participant in many PWSA meetings. While he has decided to "move over" and let some more junior scientists participate in the SAB, Dr. Hall remains very active in the Kentucky PWSA chapter and still takes care of many people affected with PWS.

Louise Greenswag, R.N., Ph.D., a nurse and psychologist, has been a member of the SAB for several years as well. She is very actively involved in helping individuals, families, and group homes with PWS issues, as well as in conducting research on issues related to behavior management. She is known to many as the co-editor of *Management of Prader-Willi Syndrome*, the book that so many of us use to improve the quality of life of people with PWS. She has chosen to resign from the SAB so that she can help PWSA in other ways.

Two new members joined the SAB this past year. They are **Merlin Butler, M.D., Ph.D.**, and **Elisabeth Dykens, Ph.D.** Dr. Butler is a physician and long-standing genetics researcher who has made many important advances in our understanding of the genetics of PWS. Dr. Dykens is a psychologist who has recently published many scientific articles on the psychological and behavioral characteristics of PWS. Both are expected to be of great benefit to PWSA, and we look forward to their contributions.

Scientific Day Report—Part III: Deletion vs. Disomy Differences in PWS

At Scientific Day in Seattle, two studies were reported that examined differences between those who have PWS due to a deletion on the paternal chromosome 15 and those who have PWS due to maternal uniparental disomy (two chromosome 15s from the mother and none from the father). Following are summaries of these two studies.

Few phenotypic differences between patients with Prader-Willi syndrome due to deletion 15q and uniparental disomy 15. *Research by S.B. Cassidy, M. Forsythe, S. Heeger, R. Nicholls, and S. Schwartz; Department of Genetics, Center for Human Genetics, Case Western Reserve University and University Hospitals of Cleveland, Ohio.*

In this study, the charts of 36 patients with the chromosome 15 deletion and 18 patients with uniparental disomy (UPD) were compared on a number of characteristics. The researchers found statistically significant differences in six areas: Those with a deletion were more likely than those in the disomy group to have hypopigmentation and "typical facies" and to exhibit skin picking, articulation abnormalities, high pain threshold, and skill with puzzles.

The researchers found no statistically significant differences between the two groups in the frequencies of neonatal hypotonia or need for NG tube feeding, behavioral disorder requiring intervention, cryptorchidism, hypoplastic genitalia, hyperphagia, sleep disturbance, viscous saliva, small hands or feet, scoliosis or kyphosis, diabetes, dental abnormalities, age at walking, mean IQ, or mean height by sex.

Although recent studies of Angelman syndrome, which is the genetic opposite of PWS (caused by either a deletion on the maternal chromosome 15 or by paternal disomy) suggests that AS patients with UPD had a "milder phenotype," this study "suggests that ... PWS due to disomy may not be milder than that due to deletion," since observed differences are few.

A comparison of phenotype in patients with Prader-Willi syndrome (PWS) resulting from interstitial deletion and uniparental disomy. *Research by J. Mitchell¹, S. Langlois¹, G. Gillissen-Kaesbach², B. Horsthemke², R. Michaelis³, A. Schinzel⁴, and W.P. Robinson^{1,4}.*

¹Dept. of Medical Genetics, University of British Columbia, Vancouver, Canada; ²Institut für Humangenetik, Universitätsklinikum Essen, Germany; ³Greenwood Genetic Center, Greenwood, S.C.; ⁴Institut für Medizinische Genetik der Univ. Zürich, Switzerland.

This study investigated clinical presentation of 80 PWS patients with maternal uniparental disomy and compared them with 43 PWS patients who have chromosome 15 deletions. The researchers found statistically significant differences in two characteristics: Parents of disomic children were older, and disomic babies were born on average 2.2 weeks earlier than the deletion babies. In addition, "results approaching significance indicated that disomic PWS children were reliant on gavage feeding for a shorter length of time ... and their onset of hyperphagia was later" than the children with deletions.

New Study on Social Relationships

A research team in Massachusetts seeks children with Prader-Willi syndrome, ages 4 to 17, to participate in a study of social understanding and social relationships in children and adolescents with developmental disorders. Funded by a grant from the National Institutes of Health, the project is being conducted by Dr. Helen Tager-Flusberg of the University of Massachusetts psychology department and Dr. Karen Levine of Spaulding Rehabilitation Center and Children's Hospital Boston and their collaborators.

In a series of home visits, the participants will be presented with "a range of interesting and fun tasks that tap their knowledge of people" and given standardized tests of cognitive ability and language. Participants' parents will be interviewed about their child's social relationships, friendships, and interests.

The researchers expect the project to "provide us with important information about the social strengths and weaknesses that are characteristic of Prader-Willi syndrome," and "lead to the development of specific strategies designed to help foster stronger social skills in children and adolescents" with PWS.

Families interested in participating in this study should contact: Dr. Helen Tager-Flusberg, Dept. of Psychology, University of Massachusetts, 100 Morrissey Boulevard, Boston, MA 02125-3393. Tel.: 617-287-6344; Fax: 617-287-6336.

Midwest Growth Hormone Study Seeks Additional Participants

The joint study of growth hormone and its effects in PWS, announced by researchers at St. Louis University and the University of Wisconsin, has openings for additional participants at both research sites. Participants must be between 5 and 18 years of age, with priority given to prepubertal children.

The study will begin with a 4-to-6-month observation of linear growth without GH treatment. Participants then will be randomly assigned to two treatment groups: the first (two-thirds of the total participants) will receive GH injections for a two-year period; the second group (the control group) will receive no injections of any kind the first year, but will have the opportunity for GH treatment after the first year. Both groups will be given regular assessments during the entire study period.

If you want your child to receive GH and think he/she may be eligible, it's not too late. Contact the research team nearest you: *Missouri*—Barbara Whitman, Ph.D., or Sue Myers, M.D., Tel.: 314-577-5600, ext. 2443 or 3244; *Wisconsin*—David Allen, M.D., or Aaron Carrel, M.D., Tel.: 608-263-5835.

The researchers found no significant differences in any of the following characteristics: reduced intrauterine movement, birth length and weight, neonatal hypotonia, gavage feeding, hypogonadism, weight and height on last examination, strabismus, and onset of walking.

Therapists from the University of Washington covered some of the basics of development in PWS at last summer's conference in Seattle. Their points are summarized for Gathered View readers in the following two articles.

Speech and Language Issues in Prader-Willi Syndrome

by Cynthia Branson, M.A., CCC-SLP, University of Washington

Children with Prader-Willi syndrome present a number of speech and language features that are important to understand to ensure timely and appropriate interventions. This article describes the most common problems seen in PWS.

Children with PWS are often first connected with a Speech-Language Pathologist (SLP) in relation to early feeding problems. The problems of low tone and poor oral motor control, first seen as a health issue, may continue and have influence on the acquisition of the speech sounds of the language. Review of the literature shows that most children with PWS have articulation difficulty that may well continue throughout adolescence.

Intelligible speech requires rapid and precise movements of the lips, tongue, and soft palate in complicated sequences. A child with oral motor problems may have more difficulty performing discrete speech-related movements like lifting the tongue tip and more problems coordinating the articulatory gestures demanded by connected speech. Moreover, when the timing of muscle movements is off, problems with resonance may occur. Weak and poorly timed posterior movement of the soft palate can contribute to the inconsistent hypernasality and nasal air emission frequently seen in the PWS group. The term *dysarthria* may be used to describe your child's speech as it pertains to weak, imprecise, and poorly timed articulatory movements. The good news is that the prognosis for improving articulation is relatively good. Even older kids with persistent misarticulations have been shown to be stimutable for improved speech sound production skills.

In some kids with PWS, the articulation problem is of a different stripe. A subset of children show speech characteristics indicative of *developmental apraxia of speech*. This is a problem of planning motor movements for speech. Stated another way, kids with this type of articulation difficulty have trouble figuring out what to tell their muscles to do to produce sounds and sequences of movements. A typical profile shows a small sound repertoire, difficult-to-understand speech, frequent omissions of sounds and syllables, problems imitating speech, groping behavior and expressive language problems. Moreover, errors may be inconsistent, with some words articulated

well and other words containing the same sounds beyond the reach of the child. In some cases speech is so unintelligible that alternative communication modes are developed, such as manual sign language or picture exchange systems to mitigate the serious frustration around communication breakdowns.

Delayed language development is common in the PWS group, although the language profiles of the group vary. In one study, slightly more than half of the children showed language skills at a level commensurate with cognitive skills. Areas of deficit often include vocabulary limitations and problems understanding and producing complex grammatical elements. Older kids differ most from peers in terms of the level of abstraction of information understood and produced.

Another subset of children with PWS show uneven language profiles with comprehension skills in advance of expressive skills. Articulation problems enter the mix in that some children with PWS do not have the speech sound production skills to produce the complicated sequences of consonants that result when adding grammatical forms to sentence elements such as nouns and verbs. Word recall problems have also been described in some children with PWS.

While language may be slow to develop, young children with PWS do gain preverbal communication skills that permit them to begin to negotiate the social arena. They learn to attend jointly with caregivers to aspects of their environment by gazing and manipulation of materials. What these communication partners share becomes the stuff of conversation later. Through social play and imitation, kids with PWS also learn to engage in nonverbal reciprocal activity, which serves as a building block for conversational turn taking. They learn, too, to anticipate sequences of events, gaining skills in waiting and in understanding how events relate to one another. They also gain a repertoire of conventional gestures like giving, pointing, and showing of objects that serve to call attention, request, and, sometimes, protest.

Clearly, communication is going on between kids with PWS and their families before speech is used as a social vehicle. For parents of children whose speech and language skills are slow to develop, it is especially important that they learn to



identify and optimally respond to these early communication behaviors they can expect to see. A speech/language pathologist can be valuable even for the child who is not talking. Through working with parents, early communication milestones can be practiced and expanded.

In the area of fluency, some children with PWS are disfluent but do not fit the clinical picture of stutterers.

While there are some common features seen in the speech and language characteristics of children with PWS, there are

many individual differences in the population that argue for individually tailored intervention programs. Parents can best advocate for their children by specifically querying school personnel and health care professionals about their experience with speech and language problems seen in this clinical group; by requesting SLP consultation early in the treatment plans for young children; and by serving as a resource to school personnel and others involved in their children's care if staff and service providers are unfamiliar with specific communication behaviors associated with the syndrome. □

Motor Development in Young Children with PWS ... Occupational Therapy as a Resource to Families

*by Kay Kopp, OTR/L and Jennifer Shull, OTR/L
University of Washington*

Hypotonia, or low muscle tone, is common among individuals with Prader-Willi syndrome. Low muscle tone affects the efficiency of muscle action and motor control.

In the early months of life, this is a factor in feeding. An infant with PWS may have a weak suck and have trouble feeding. Low muscle tone of the cheeks, jaw, and tongue may be contributing to this. The infant may have difficulty forming a seal around a nipple for adequate sucking. Further problems may arise with the introduction of solid foods. An occupational therapist (O.T.) can help families with feeding in several ways, from suggesting an adapted nipple, to using a variety of positioning and handling adaptations, to changing the texture of the food. An O.T. can be consulted at any time for help with feeding, from early infancy on.

As the child gets older, low muscle tone may impact postural control for independent sitting and early mobility. For example, children with PWS may be late in taking their first steps. Relatedly, balance, coordination, and fine motor skills may also lag behind. An O.T. can help the child and family to encourage normal movement patterns as well as develop sensorimotor skills (the combination of sensory and motor activities) that the child needs to be more independent in play, self-care, and learning at home and school. An O.T. will often be part of the child's educational team in early intervention programs and then in the public schools.

Children with PWS often avoid physical activities because they are difficult for them and require extra

energy because of the reduced muscle tone. Inactivity is a major problem for these children, particularly as weight control becomes an issue. The best advice for families is to start early in making regular physical activity a part of the family routine. There are benefits for all family members, but particularly the child with PWS. Physical activity helps improve muscle tone, aids in weight management, and promotes general motor development.

Suggested activities include:

- visits to the playground
- family walks
- swimming classes
- toddler tumbling
- movement classes
- children's exercise videos
- dancing to music
- bicycle riding and pedal toys
- recreational activities in the community (Check your local parks and recreation department.).

Parental example of regular exercise and physical activity serves as a role model to pattern positive lifelong habits for their children. It is important to start early and be consistent. Make physical play a part of each day!



My Brother, My Hero

An Address to the Siblings' Workshop at the 1995 PWSA Conference

by Shannon MacDonald, Redmond, Washington

Hi, my name is Shannon MacDonald. I am 19 years old and have been living with my brother, Craig, for 18 years. Craig has Prader-Willi syndrome.

Being a sibling to a brother with PWS comes with many challenges. I have had to stand up countless times to other kids who were teasing my brother. My philosophy is, I am the only one who is allowed to tease Craig. If anyone else does, they are dead meat, no matter what their size. I have had to deal with him stealing and breaking my things. It has finally gotten to the point that I lock my room to keep my possessions safe. I have had to apologize or explain to people why he took something from their store, garage, or home. I have had to give up my weekends with my friends to baby-sit Craig so my parents could have a break. At times it has been very hard, but with a brother with PWS I usually try to accept things for the way they are. However, I do get angry and frustrated about the way things have to be.

I remember when I was younger and spent the night at a friend's for the first time. I noticed their refrigerator and cabinets were not locked. I realized then that my life was very different from my friend's.

Living with Craig has been very interesting. My parents and I feel we could write a book on all our experiences with my brother. Each day there is a new story. It's

gotten to the point when he does something wrong we get upset, but later we are able to laugh about it.

I will always be Craig's sister. There is no changing that fact. Yes, he is frustrating, challenging, tiring, unfair, and at times embarrassing—but life with Craig can be very rewarding. My brother has taught me many things. I am so proud of him and all he has achieved because I know he has to work twice as hard as others just to reach a goal that most of us would consider simple. He is graduating from high school next year, and I have never been so proud of someone.

When people ask me who my hero is I don't have to think. I just simply say, "Craig, my brother," because he has taught me to be courageous. I see him struggling daily with his syndrome, kids teasing him, people constantly watching his every move as if he were a prisoner, but he keeps on going. He deals with things every day that would have driven me crazy a long time ago. Craig has taught me how to be forgiving and loving. No matter how angry he gets, later Craig will say "Shannon, I am sorry," give me a big hug, and say "I love you." Craig has taught me so much about life and has given me so much. I wish that I could give him half as much in return.

In our religion we always give up something for Lent. Last year before Lent Craig and I were discussing what we would give up. Craig said, "Shannon, I want to give up Prader-Willi for Lent." How I wish I could make that wish come true.

Being a sibling to a person with PWS is not just about locking things up or about anger and frustration. It is about being proud of someone, loving someone for what he is, making sacrifices for him, and trying to achieve the courage he has. It is about standing back and admiring him and the things he has achieved. Most important, it is about making him my hero.

Ask the Professionals

cholesterol

Q ■ My daughter, who is 25, just had a physical exam with accompanying blood tests. Her cholesterol was 294, which is very high. Is high cholesterol associated with Prader-Willi syndrome?

A ■ Cholesterol abnormalities are not an intrinsic part of PWS, as far as we know. Those affected individuals who are sticking to a low-fat diet and who have no family history of high cholesterol levels are not more likely to have elevated cholesterol levels than anyone else in the population. However, they are not protected from high cholesterol, either. If they ate a high-cholesterol meal (or snack) before the test, if they consistently eat a high-fat diet, if they get little or no exercise, or if there is a gene in the family for high cholesterol, they are as likely as anyone to have high blood cholesterol. Your daughter's high cholesterol should be investigated like anyone else's. (Suzanne Cassidy, M.D., Case Western Reserve University)

Portrait of a Lady ...

a Mother's Reflections

by Sarah Abell
Louisville, Kentucky

She came into the world on September 8, 1967, following a relatively uneventful pregnancy. I had expressed a concern about the absence of fetal movement. The doctor dismissed it casually, saying, "You'll probably have a mild mannered little girl."

Our "mild mannered" little girl was an unusually beautiful baby, dispelling the anxiety that had accompanied my concerns about diminished fetal activity. We named her Joyce Lynn. It was evident at the hospital that she was not interested in the formula, sleeping most of the time. No problem; I could handle that when we were at home. So we went home, and in no time feeding her became a real challenge. It required hours to force a few drops down her. Weight gain was slow. The pediatrician recommended a change in the formula to enhance its caloric value. He maintained that she would "catch up." Eating improved toward the end of the first year, and so did the muscle tone.

The youngest of 15 children, Joyce began to thrive at the heart of her loving family. Though developmental milestones were somewhat delayed, she was happy and had many role models and loyal playmates. Her early childhood was not markedly different from other children her age.

We were introduced to the term "Prader-Willi syndrome" at a follow-up evaluation when Joyce was 8. (She was chubby but has never been obese.) No information was offered, and our research in 1975 revealed very few resources. A short time later, I came upon an obscure little notice in a publication that an organization was being formed in the interest of Prader-Willi syndrome. Membership in the Prader-Willi Syndrome Association (USA) has been a godsend. The association became the source of helpful information as we began to deal with the food seeking and other behaviors. Months of counseling had produced very little results.

Then came the school years. Joyce arrived on the education scene at the time of special education in self-contained classrooms with some mainstreaming. She benefited to the extent that she acquired good reading skills and rudimentary



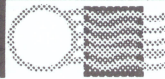
math, making the most of her abilities.

High school years were marked by several events—some good, some not so good. Her father passed away; she underwent surgery for scoliosis; her childhood friends drifted away. The prom was the highlight of her senior year. She attended in a manner that could be the envy of any young lady. Her date and boyfriend was a handsome classmate, quite debonaire. Joyce was stunning. Her sisters applied all the beauty techniques of style and makeup.

Next thing we knew, Joyce had graduated only to find that there were no opportunities for a fulfilling adult life style. It was after years of perseverance and diligence by the Prader-Willi Kentucky Association, and later, ARC of the Blue Grass, that a Prader-Willi home was established in Lexington. Castlewood opened in early 1991 and is the home of eight individuals from Kentucky who have the syndrome. The residents have work, activities, and opportunities for socialization that could never be duplicated with family resources. The program has been successfully orchestrated by the leadership of the two organizations.

Joyce knows all about her syndrome and clings to the hope that doctors will somehow find the solution to why people with PWS can't have babies. She is very anxious to participate in Dr. Butler's research project. Joyce likes people, babies, pets, and nice-smelling cosmetics. She particularly likes sending greeting cards and letters to family and friends.

Joyce comes home frequently and calls often. Our telephone conversations always include glowing accounts of the meals at Castlewood. She has assumed a measure of independence. She now makes some decisions for herself and feels good about it. We have come to terms with the separation. She is missed at home, and it has been difficult to relinquish part of the role of caregiver. We are grateful to the manager and staff of Castlewood for their caring and dedication. We look forward to an even brighter future as more of the secrets of PWS are unfolded. □



A Lesson in Hunger

Greetings from Japan! My husband is in the U.S. Navy and so we are currently living in the Land of the Rising Sun. We are PWSA members by virtue of the fact that our Hannah (age 14) has PWS. I have benefited greatly by receiving *The Gathered View* over the years. I have not always enjoyed what I've read, but have learned (I'm sure you know what I mean).

I wanted to tell you how we participated in PWS Awareness Day. About that same time, the youth group where Hannah is a member sponsored (with World Vision) a 30-hour fast. It was intended to raise hunger awareness for poor countries and the kids raised money to send to one of the poor countries. Hannah badly wanted to join the fast. I was skeptical, but her motivation and perseverance won, and we agreed that she would fast for 24 hours. We set up a liquid chart so she could drink regularly and know when the next drink was coming. She was successful in fasting from 7 p.m. one day to 7 p.m. the next and raised over \$100 for kids less fortunate than herself. (But that's not even the punchline of the story!)

About halfway through the fast I went to the place where the kids were gathered (Hannah was at home). I asked the teens, in greeting, "How are you?" To a person, each one said, "HUNGRY!" I then went on to share with them that no matter how much or how little Hannah eats, she feels the same way, all the time. I was then able to share some details of her syndrome and some ways they could understand her better. It was a very effective means of making my small world aware of Prader-Willi syndrome.

Keep up all the good work for our PW guys ...

*Kathy Switzer
Japan*

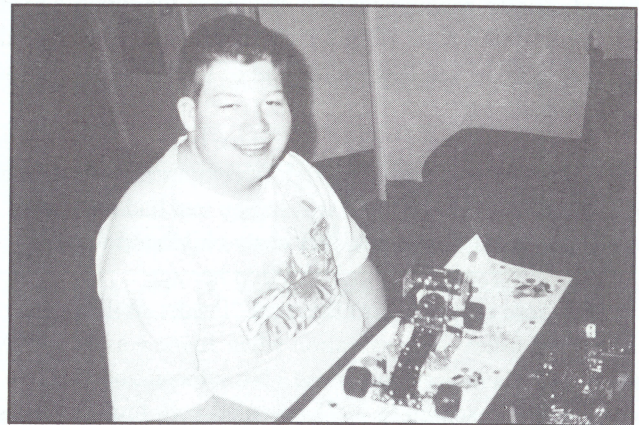
Editor's note: Kathy and her family plan to return to the United States soon and settle in Columbus, Ohio. Through the national PWSA office, they've been put in touch with the Ohio chapter for help in locating a school program for Hannah.

Spreading the Word in South Dakota

Karl Jacob (K.J.) Schauer, of Dupree, South Dakota, was chosen as the 1995-96 South Dakota Easter Seal Child and honored at an Easter Seal Society awards banquet and breakfast last September in Rapid City. Among his duties, K.J. will represent the Easter Seal Society at the state Legislature in Pierre in January.

K.J.'s parents, Carl and Velma Schauer, sent PWSA a newspaper clipping from the Dupree *Progress* and wrote: "We felt this was a real honor and also helped educate more people about Prader-Willi syndrome ... If this can encourage other children or parents dealing with PWS, this would be great."

K.J. was born on Dec. 24, 1979, and diagnosed with PWS at the age of 14 months. Currently, he is a sophomore, mainstreamed in nearly all of his high school classes. K.J. plans to attend a training workshop after high school and "hopes to become a preacher, farmer, construction engineer, or a teacher, marry and adopt many orphan children," according to his newspaper interview. Carl and Velma told the reporter that K.J. loves to visit with people of all ages, tell jokes, and play with animals, and that his favorite hobbies are collecting antique toy tractors and vehicles, putting puzzles together, and erecting Steel-Tec sets.



K.J. Schauer with work in progress

The national PWSA office is in the process of updating its database of local resources for persons with Prader-Willi syndrome. Chapter presidents have been asked to update resource lists for their states or regions and to work with the national office to keep them current.

If you know of experienced professionals or groups providing services to those with PWS (doctors, therapists, schools, group home providers, rehabilitation clinics, vocational programs, camps, etc.), please call, mail, or fax the information to your chapter president, or in areas with no chapter to the national PWSA office, as soon as possible. The right name and phone number can make a world of difference to a family in need of services.

A Report from the 1995 NORD Conference

by Judy Livny, Parent

On September 30 and October 1, I attended the NORD (National Organization for Rare Disorders) Annual Membership Conference in Iselin, New Jersey, as a representative of the New Jersey PWSA. The conference was cosponsored by *Exceptional Parent Magazine*.

Along with approximately 300 other attendees from all over the United States and a few from Europe, I chose several workshops to attend. A sampling of the topics discussed is: Genetics 101, Ethical Issues of Genetic Research, Fundraising for the Non-Profit, Lessons in Lobbying and Advocacy, and How to Motivate Volunteers and Develop Chapters.

A special presentation on orphan diseases and drugs in Europe was made by Lesley and Peter Greene of England, the founders of RTMDC (Research Trust for Metabolic Diseases in Children). Other special addresses were given by FDA Directors Marlene Haffner, M.D., Office of Orphan Products Development, and Philip Noguchi, M.D., Division of Cellular and Gene Therapies; by Stephen Groft, Pharm.D, Director of the National Institutes of Health (NIH) Office of Rare Disease Research; and by Stanley Klein, Ph.D., Editor-in-Chief of *Exceptional Parent*.

As always in conferences like these, one of the highlights is the people you meet. The variety of disabilities represented was mind-boggling. NORD offers literature on approximately 500 different diseases and syndromes [including PWS].

Though all the sessions I attended were interesting, I particularly enjoyed Joseph Gatto and Steve Friedlander's presentation on Fundraising for the Non-Profit.

Here is a short quiz to see what you know about charitable giving:

1. How much money was contributed by Americans from all sources last year?
 - a. \$20 billion
 - b. \$50 billion
 - c. \$75 billion
 - d. None of the above

(The answer is d. Last year Americans donated \$130 billion to nonprofit causes!)

2. What was the largest source of contributors?
 - a. Individuals
 - b. Corporations
 - c. Foundations
 - d. Bequests

(The answer is a. Individuals contributed about 88 percent of \$130 billion.)

3. Charitable giving as a percent of income is highest among?
 - a. High-income households
 - b. Low-income
 - c. Middle-income

(Low-income families give the most, percentage-wise. Middle income households give the least.)

Resource Shelf



Two new publications are available in the *Prader-Willi Syndrome Information Series*, available from *Prader-Willi Perspectives*:

The Child With Prader-Willi Syndrome: Birth to Three (PWS Information Series, No. 3), by Robert H. Wharton, M.D., Karen Levine, Ph.D., Maria Fragala, P.T., and Deirdre C. Mulcahy, M.S., CCC-SLP, at the Center for Prader-Willi Syndrome & Related Disorders, Spaulding Rehabilitation Hospital, Boston, Mass. Price: \$5.00 per copy for up to 4 copies; \$4.00 each for 5 to 9 copies; \$3.00 each for 10 or more.

Prader-Willi Syndrome: Information for Parents and Professionals (PWS Information Series, No. 4), Merlin G. Butler, M.D., Ph.D., Vanderbilt University College of Medicine, Nashville, Tenn. Price: \$7.50 per copy for up to 4 copies; \$7.00 each for 5 to 9 copies; \$6.50 each for 10 or more.

A 1995 revision of the first booklet in this series, ***Children With Prader-Willi Syndrome: Information for School Staff***, by Drs. Wharton and Levine of the Spaulding Rehabilitation Hospital in Boston, also is available from the same source. Pricing is the same as for No. 3 in the series, above.

Orders and payments for the above booklets should be directed to: Visible Ink Incorporated, 40 Holly Lane, Roslyn Heights, NY 11577-9890.



Fat-Free Holiday Recipes, by Sandra Woodruff, R.D., Avery Publishing Group, New York, 1995. Paperback (251 pages), \$13.95.

Here's a cookbook that's useful year-round, in addition to helping dieters survive the holidays. Making use of nonfat dairy products, dressings, etc., the author overhauls popular hors d'oeuvres, desserts, and treats and offers tempting menus for favorite celebrations—Thanksgiving, Christmas, Hanukkah, Passover, Easter, New Year's Day, Valentine's Day, St. Patrick's Day, Halloween, Memorial Day, Fourth of July, and Labor Day. This attractively designed book includes an informative first section entitled "Getting the Fat Out of Parties," color photographs, nutritional facts for each recipe, a products resource list, and an index.

Do you have a story or experience you'd like to share with other families? —a concern or question you'd like to pose to parents or professionals? —a great recipe to share with other families or group homes? —a resource or service you think others should know about?

If so, let us hear from you!

Contact the national PWSA office by mail, phone, fax, or e-mail—anytime.

Our Deepest Thanks for Your Gifts

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In honor of Gladys Wolf's 85th birthday— Denny Singer

In honor of a special, giggly granddaughter, Stephanie Baker—Mr. & Mrs. Richard P. Smith

The Answer is Yes

- Can persons with Prader-Willi syndrome learn and benefit from a good education?
- Can persons with PWS have friends?
- Can persons with PWS have special aptitudes and interests?
- Can persons with PWS get and stay physically fit?
- Can persons with PWS grow up and move out of the house to a suitable adult living situation?
- Can persons with PWS get a job?
- Can families of those with PWS have a rich and happy family life?
- Can living with PWS be challenging to your heart, your mind, and your soul?

(reprinted from *Prader-Willi Voice*, the Ohio PWSA chapter newsletter)



Support Systems

My right hand is being held
by someone who knows more
than I, and I am learning.
My left hand is being held
by someone who knows less
than I, and I am teaching.
Both my hands need thus be held
for me—to be.

"Is There Where I Was Going?"
by Natasha Josefowitz

(reprinted from Iowa Exceptional Parent
Center's *Special Edition*)

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

