

The Gathered View

Newsletter of the Prader-Willi Syndrome Association

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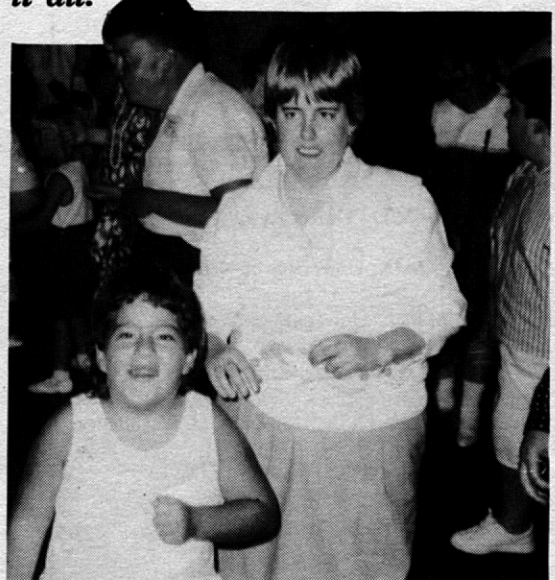
Prader-Willi Syndrome Association Conference "now history"

As Dr. Beltran writes in his "President's Message," (see page two) the 11th Annual conference is "now history." We are happy to report that attendance followed fairly close to our usual attendance with 248 registered adult attendees and 101 in our activity program. Comments made and evaluation sheets shared gives us the impression that efforts were worthwhile and we held another successful conference. We have asked a "first time attendee" to write his impressions of the meeting, which is included in this issue along with some photos that we are happy to share. The evaluations always share that the opportunity to meet and talk with other parents is the greatest part of the planning. We are all looking forward to our next conference in July, 1990 in Salt Lake City.

Door prizes were offered at the conference to assist in supplementing the conference budget. The round trip airline ticket to anywhere in the U.S. was won by Dr. P. Wilkinson of Lethbridge, Alberta, and his wife, Joyce won the 2nd prize of one of our autographed, numbered, Management books. The 3rd prize, another book, was won by Rhett Eleazer of South Carolina.

One of the board actions was accepting the bid from the Illinois chapter to host the conference in 1991. Some confusion has developed because of the plans being made to hold a meeting in the Netherlands. We would like to clarify the fact the 13th Annual Conference on PWS will be held in the Chicago area. The Netherlands meeting, which is still very much in the planning stage, will be financially sponsored by PWSA but this meeting will not take the place of our usual annual conference. Further information will be shared with the membership as plans develop. At the present time, Dr. Cassidy and committee are working with Henk Moezelaar of the Netherlands and Jean Phillips-Martinson of Sweden in the development of this meeting.

A window on the conference . . . their smiles say it all!



PWSA

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President's Message:

The Eleventh Annual PWSA Conference was held in Calgary, Alberta, Canada this year and is now history. Each year the conference demonstrates the maturing nature of the organization. The Scientific Sessions have developed into reports of fundamental research as well as the reports of observations of the Prader-Willi individual's characteristics. The main meeting provided a look into the social aspects of the lives of individuals with PW that had not previously been touched. But it was the meeting of the Chapters leaders that brought to my attention some changing aspects of organization that cause me concern.

The lifeblood of any organization and the reason for its existence is the people who want to be identified as members of the organization. It is stated clearly in the By-laws that the PWSA exists for the improvement of the lives of people with Prader-Willi syndrome. The goal of increasing awareness and educating the public and professions is being met on a grand scale. This is easily demonstrated by the fact that the new text edited by Director Louise Greenswag and her associates has gone into a second printing. It is also demonstrated by the decreasing number of state legislators that do not know of the syndrome.

The lives of many persons with Prader-Willi has been greatly improved by the development of over twenty dedicated residential facilities. There are more and more reports of earlier diagnoses of persons with the syndrome, evidence that the medical community is more aware, largely due to the efforts of the association and its membership.

Why then did our chapter presidents report a large disaffection for supporting local chapter efforts. Almost every report indicated that meetings were attended by the same few individuals that held the waning group together. That there were parents who would not make the effort to help in the efforts of the group to raise funds, or engage in projects, or to accept the responsibility of holding an office. Even more devastating is the unwillingness to participate in voting for officers and directors of the board for the local as well as for the national meeting. This is not unlike our state and national political elections where fewer and fewer people partake of the fundamental right of a democracy, to elect their own representatives.

When apathy at any level of organization is evidenced by indifference of the voting membership the organization can be threatened by various forms of destructive energy ranging from corruption of power to insensibility. An even more subtle destructive force that can develop is evidenced by loss of communication between the members and the development of factions fostered on rumors and innuendo. These are unpleasant thoughts and no more easy to write

(continued on Page Eleven)

Banquet Honorees

A few years ago PWSA started a tradition of honoring people during our conference banquet. Naturally, the first people thought of were those instrumental in founding the organization. Seeking information on PWS, the Deterlings journeyed to Seattle, WA. It was there, with the help of Dr. Holm, staff, and a few other parents, that PWSA was founded in 1975. We were happy in 1984 to be able to honor Gene Deterling as our first president and Shirley Neason as our first newsletter editor. (Fausta Deterling certainly deserved "honorable mention" also as a founder.)

The next year we chose Dr. Vanja Holm, who Dr. Beltran lovingly refers to as our "Mother Superior." Her establishment of the PW clinic in Seattle, working and caring all these many years, has benefitted us greatly.

In 1986 I felt it a privilege to award our thank you plaque to a dear friend, Dorothy Thompson. Dorothy's story is that two people with PW in her 103-bed facility took up 90% of her staff's time. It was then she decided these people needed help. Luckily she has been giving that help ever since through our organization. To the list of founders and Mother Superior, we add "Guardian Angel."

In 1987 the tables were turned on me--the person scheduled to be honored by usurped by D.J. Miller and with the help of many others surprised me with that honor. I have a beautiful memory book containing letters from our membership that is one of the most treasured gifts I have ever received. The following year we followed through with the plans and did honor Dr. Delfin Beltran. Sam, as he is better known, has been involved in our group since the first parent-professional meeting in Washington in 1976. He served as one of our first board members, as Board Chairman, and stepped into the role of President many years back. His many contributions to PWSA have contributed to our successful growth.

This year I chose to embarrassingly keep this honor "in the family." To ignore the fact that the person being honored is my husband and to place his name on this deserving people list is done because he is just that--he is deserving of this honor. Dick was the first replacement on the original Board of Directors, he served many years as Chairman and still remains on the board. In 1979 he willingly gave up a room in his home for the establishment of an office for the national headquarters and the office remained there until a year ago. He has always been my right hand when he was needed. Particularly in the past few years, he has been indispensable as our computer consultant. He has taken over the duties of running the GV labels, membership listings, and all of the many things we previously paid hundreds of dollars to have done. We also have another computer consultant, Loren Goff, Judy's husband. Without the help of Dick and Loren many of these money and hour saving innovations would not be operating.

But most of all, I believe Dick deserves this honor because he is always there to listen. A person could not remain in my job for the past ten years without a sounding board. It makes it a lot easier to work through problems when you have someone to share them with. Luckily, the rewards of this job far exceed the problems, and some of that can be contributed to the many good people who are members of the organization. So we honored Dick at the banquet and at the same time said thank you to the rest of you also.

Calgary has a special ceremony to honor people and all of the attendees enjoyed becoming "Honorary Calgarians." Dick's certificate stated, "This here paper proves Dr. Richard Wett had the purely pleasurable experience of visiting the only genuine cowtown in Canada...namely Calgary, and was duly exposed to exceptional amounts of 'heart warmin', 'hand shakin', 'tongue-loosinin', 'back-slappin', 'neighbor-lovin', western spirit...is hereby deputized as an honorary CALGARY WHITE HATTER said title means from this day on, the owner of this certificate accepts responsibility for extendin' this brand of western hospitality to all folks and critters."

-- Marge A. Wett



A Letter to the Prader-Willi Syndrome Association

The illness and death of our darling daughter, Lynnel this past February has left me with an urge to notify other parents of certain signs and symptoms that they may not be any more aware of than I was. She was diagnosed with PWS at age 8. Her symptoms were primarily classic. She was on as good a diet as can be maintained with a child living at home, semi-independent and very active. She was in a hospital for a month at age 18 for a fasting diet. This worked pretty good on 500 calories a day. However, this was not encouraged at home with all her numerous physical activities. Up until her years' illness, her weight remained around 180-210. I'm sure she was 250 lbs. at the time of her death. Although her calories were watched closer then, all her physical activities were stopped (sheltered workshop, swimming, recreational activities), busy every minute of every day. Naturally as she became less active her weight increased. Walking was the only activity she was allowed, and this had to be carefully monitored. She began having respiratory problems in April, 1988. She could not breath lying down -- she had to sit up or be elevated on pillows. After 10 days of antibiotics, it did not improve and she was hospitalized. She also began a menstrual period two days prior to hospitalization which she had never before had. Immediately upon hospitalization, hypoxia was suggested and confirmed and oxygen administered along with lasix, theophylline and an antibiotic. Pneumonia was guessed but, due to the inability to get a clear chest x-ray was never confirmed. An enlarged right side of the heart indicating congestive heart failure was confirmed. Menstrual bleeding was confirmed and she was started on a hormone therapy to regulate the periods and possibly help with the fluid retention. The therapy worked to a degree but she began bleeding every time she became congested throughout her illness no matter when it was. My theory was that this was the body's way of attempting to rid itself of excessive fluid buildup.

After three weeks, it was decided no more could be done. This was after every test conceivable had been administered. She was brought home and was on an in-home program of 3-5 liters of oxygen 24 hrs. per day plus theophylline and lasix. These were regulated by visiting nurses until the theophylline could be adjusted to her system. Unfortunately, the bouts of respiratory distress became more frequent and meds had to be continuously increased as did the oxygen. She died February 12th peacefully at home with us. She was 24 yrs. old.

My suggestion to other parents is to watch for signs of hypoxia! Lynnel periodically turned blue

(her lips, her fingertips, her toes) all through her life usually during times of excitement or stress. This was never mentioned to me as her possibly being hypoxic, and it was so very difficult for anyone to draw blood that they seldom did when she was in the hospital or just to see a doctor. Fortunately, during this last hospital stay, there was a doctor there who could draw blood and they did numerous tests by being able to do this. I firmly believe she had a degree of hypoxia all her life—even before she became so heavy. The doctors stressed the fact that a person's body usually always maintains the proper amount of oxygen no matter what their size is, and they never figured out the problem as her lungs seemed clear. Also, there is an overall deterioration of the lungs due to PWS children always being very shallow breathers. Lynnel was never capable of taking a deep breath as a normal person is, and this caused the lungs to atrophy over the years. Lynnel was also confirmed as having sleep apnea several years ago, and the test was repeated in 1988 and confirmed that the sleep apnea had worsened. The doctors also suggested this really has nothing to do with a person's weight. Although, through the years, it certainly has something to do with the oxygen saturation in the body.

Lynnel was first on a waiting list to go into a PWS home three years ago which was just three miles from our home. Naturally, after she became sick, it was too late. Lynnel was an only child, her step-father is a paramedic, she was an extremely happy-go-lucky young adult, always had a smile and never had the temper tantrums that are one of the PWS symptoms. Consequently, having her at home was a joy for us. However, when she was encouraged by her school to go into the PWS home and was all for it, we went along all the way. Unfortunately, she was not chosen at that time.

In summary, parents must be more aware in the signs and symptoms mentioned above. When the PWS child goes into heart failure, it comes very rapidly. Up until the Thursday night before Lynnel's death on Saturday night, she was not in enough distress to really be noticeable. She kept her smiley personality and never once complained right up to her final breath. Her optimism throughout her lifetime would put normal people to shame. She never complained about all her activities being curtailed--when she got better, she would resume them. Their positive attitudes are as strong as their "angelic qualities" in securing food from strangers.

Perhaps more research should be done concerning the oxygen saturation level, the sleep apnea and the shallow breathing in PWS children.

A "First-Timer" Comments on the Conference

It's a long way from New York to Calgary. It's a long way both in terms of miles flown and years passed since the diagnosis of PWS hit our family like a locomotive. I'm on my way to attend my first national conference. On the plane the mind begins to play those tricks for which the human mind is (in)famous -- fear, anxiety, dread, apprehension, and just about any other synonym my thesaurus can come up with. The fear, etc., is tempered with a feeling that I'm doing a very positive thing by attending this conference. Maybe some of the fear can be assuaged through the learning process. *Maybe I'll find answers -- if not answers, maybe I can begin to better understand my questions!*

The plane lands. *Reality is just a short ride away.* Arriving at the hotel I enter the lobby. In the lobby are several people with PWS. Some are young children; some are adolescents and adults. Some are obese; others have obviously been able to control their weight. *Am I looking at Emily's future?*

One thing about PW kids -- and adults -- they are friendly and will usually strike up a conversation with you right off the bat. "Are you a Prader-Willi?" one youngster asks me (I'm a bit overweight, so maybe he was fooled!). "No," is my reply, "but I have a daughter who is." "How old is she," I'm asked. "She's six," I answer. What's her name, my new friend asks. "Emily," is about all I can offer as the tears begin to ooze, even though I try hard to be brave. "Oh," is his answer. "See you later," he concludes our conversation with a sincere handshake and moves on to another adventure. *Not a bad-looking kid. Maybe Em will grow up to be like him. Wouldn't that be nice!*

The ice being broken, the initial emotions having been encountered and shelved for the time being, I can concentrate on getting down to business, getting to know my new "extended family," getting on with my life as a father of a child with Prader-Willi Syndrome. As the hours and days unfold, I reaffirm my belief that attending the conference was perhaps the most positive action I could possibly take. There's a perspective out there, and maybe I'll pull it into focus as the week goes on.

I'll not clutter up my reminiscences with the facts of the conference -- they're in my mind for keeps and will surely be recounted by others. There was much more going on in Calgary that week than the superb meetings, lectures and workshops. What I hope to convey to you, readers of this newsletter, most likely parents of PW people, is that by attending the national conference, I realized for the very first time that I am not alone. Not by any means. There is a family out there waiting to share with you, waiting to help you handle the problems, the pain, even the pleasures, eager to get in touch and stay in touch. Next year we meet again, and I will be there. In fact, I wouldn't miss it for anything. I hope more of you will be there, too. I won't be a "rookie" anymore, so maybe I can be a comfort to you just as so many fine people I met were a comfort to me. -- *Sheldon L. Tarakan, Long Island, NY*

Dear Gathered View . . .

"Just thought I would share with you a picture from our recent trip to Switzerland. We stayed three days in Zurich where the Children's Hospital is located. This is the hospital our famous Dr. Prader is located. We ventured out one morning and with a little luck and the grace of God we were able to meet him. We had a nice talk and he asked of our daughter, Beth. When we told him that she was 14 yrs. old, 56" tall and 85 lbs., he looked at us and with all sincerity he said, 'you are to be congratulated'. Dr. Prader is a very humble and gracious man, giving much credit to his partners Drs. Labhart and Willi. We thanked him for all that he has done and he stated, 'I did very little--there is so very much more to be learned and discovered.' Meeting Dr. Prader was one of the highlights of our trip, and we felt it a great honor to talk with him.

Also, July 1st, our newly formed North Carolina chapter of PWSA was honored with the presence of Dr. Jeane Hanchett from the Rehabilitation Institute of Pittsburgh. She presented a very informative talk on PWS. As always, we very much enjoyed being with Dr. Hanchett--she is at the top of the list of dedicated professionals and a wonderful human being." -- *Penny and Bob Townsend*



For Your Information

Louise Greenswag, Ph.D. and Randell Alexander, M.D.

Question: I read about precocious puberty in the Dec.'88 issue of the GV. My 7-yr. old started to develop pubic hair, oily scalp hair and blackheads around her nose. She turned 8 in 4-89 and I have noticed that the amount of her pubic hair has not increased, there has been no breast development or appearance of underarm hair. I have many concerns but I am primarily interested in knowing if she will remain shorter than the average PWS female because of the onset of this "puberty" so young. What has happened to similar cases? We will appreciate any information and will be glad to share our daughter's history to help others. Mrs. K. Costa

Answer: To better understand what you are observing in your daughter, it is important to define two terms: "true" precocious puberty and "pseudo-precocious" puberty. True precocious puberty involves an activation of a part of the brain known as the hypothalamic-pituitary-gonadal axis. When activated, this part of the brain begins cyclical release of brain hormones which in turn triggers the release of sex hormones from the ovaries or, in the case of males, the testes. This "true" precocious puberty does not fully occur in individuals with PWS.

Pseudo-precocious puberty involves physiological mechanisms other than triggering the activity of usual sex hormones. The vast majority of cases occur when the adrenal glands (which sit on top of the kidneys) release more than the average amount of a particular hormone. Curiously enough, this type of male hormone is usually produced in small quantities by both males and females. Children with pseudo-precocious puberty develop sparse pubic hair and sign of acne but do not develop further--exactly what you describe for your daughter.

No doubt you can tell that this topic can get very complicated. Pseudo-precocious puberty has been reported in individuals with PWS. Breast growth has been described which, on examination has revealed fatty tissue rather than gland development. Some instances of "menses-like" spotting has also been observed but not regular menstrual flow based on cyclical sex hormone function. Pseudo-precocious puberty does not cause abnormally advanced bone growth leading to short stature and we are not aware of any reports of a relationship between short stature and precocious puberty in association with PWS. Please keep in mind that females with PWS only average 4'8" in height because of the syndrome itself.

We suggest that your doctor check to be sure that everything is as it appears. We would be interested in following your daughter's progress over a period of time and comparing her growth and evidence of further sexual maturation to other females of her age. If you have other questions, you might contact Suzanne Cassidy, M.D. (University of Arizona Health Ctr.) who has written extensively about hormones in the PWS.

Waterwalking -- A New Possibility

Waterwalking -- an exercise that does not require special equipment or even swimming skills. Recommended for thigh to chest-deep water, 20 minutes at least three times a week. Potential benefits are the same as for walking, jogging on land, improved muscle tone, heart health, flexibility and calorie-burning (up to 460 calories per hour), but without the sweating and risk of injury. Water helps to support the body weight (approx. 90%), decreasing the trauma to the weight bearing joints. One excellent feature is that water offers resistance to movement in all directions. This means all the muscles can be exercised against resistance for a more uniform workout.

It was reported that more than 2,500 people waterwalked at the Cleveland YMCA, Norman, OK, and countless others are doing it at Y's, health clubs and in private pools. According to "Walking" magazine, at least waist-deep water, preferably chest-deep is recommended for increasing calorie burn. Swing the arms underwater to work the upper body. A long straight pool is ideal but you can walk back and forth, alternating left and right turns, or try walking backwards, sideways alternating short, quick steps with long steps or even try some fancy step kicking or knee-touching with opposite hand or elbows. Sounds good, especially for those who are overweight and exercise can threaten joint injuries.

Welcome Aboard

It is always a sad day when active board members decide not to run for re-election and in the case of Fausta and Gene Deterling it is after many many years of "sharing and caring." One consolation is that a board action established an Advisory Committee which will include the Deterlings as permanent members. The other consolation is that two terrific people were elected to take their place on the board. We are very pleased to welcome Mildred Lacy of Louisville, KY and June Smith of Enfield, CT as our newest board members, knowing both persons will do a terrific job.

Sharing

And while talking about sharing, a doctor mentioned at the conference that she would like to "write" for the GV. I do hope that all members (parents and professionals) feel free to submit letters or articles at any time for this newsletter. We welcome what you have to share and we are certain our readers do too. Naturally, we may not be able to print everything we receive or we may have to condense longer letters but please keep them coming.

Fund Raising -- a "Four Letter Word"?

Years ago a priest stated during a homily that if the pastor of a church spent all of his time begging for funds he would lose the members of the church and no longer have anyone to listen to this call for funds. That message keeps returning even though it was heard years ago.

As Director of PWSA, it is part of my responsibility to work with the treasurer and board to maintain a workable budget. In the past we have held special raffles, yearly fund drives, changed the dues when we felt it was necessary, increased conference registration fees to meet costs and in general attempted to "keep our head above water." When your organization operates on "dues and donations" that has to be a way of life.

This year I decided to purchase 1990 Pocket calendars as a means of fund raising to offset the increasing costs of running the annual conference. (This was offered to the members in the last GV issue.) We ordered 750 of these which we felt was a reasonable number considering the fact we have approximately 1500 memberships in our organization. We priced them at \$10 U.S. and \$12 Canadian (which is

retail value), and are offering an opportunity to also win cash prizes. The small trickle of orders does concern us.

These calendars can be purchased for your own use, can be sold to others as raffle chances, (one member took a dozen home and her daughter, with PW, sold them easily in the neighborhood and ordered 20 more, a special thanks Julie), can be given to others as gifts (aren't we always looking for a token way of telling someone thanks or that we are thinking of them?). We hope you as members will not let us down. We feel the conference is of great benefit to those who are able to attend but also that benefit is passed on to all of our members. Please take time to refer to your last GV and help us clear the shelf of these calendars in the near future.

And speaking of raffle chances, every calendar you purchase qualifies you for four drawings. Early purchasers qualified for an end-of-August drawing for \$50. This was won by Mrs. George Castle of Redding, CA. Three additional drawings for \$150 each will take place in January, February and March 1990. Get in on the action now!

Monetary Duties of the Prader-Willi Syndrome Association

A member recently wrote, "it should be the duty of our association to investigate and report to members the monetary programs available to them". She had found through a conversation with a friend that she could obtain funding for a home care program.

What is PWSA? PWSA is a group of parents, professionals, and interested individuals who are willing to take active responsible roles in improving the lives of children/adults with PWS and their families. This national organization, found to provide a vehicle of communication for these groups, has created and continues to maintain an international network of information, support services, and research endeavors to meet the needs of affected children/adults.

What isn't PWSA? PWSA is not a funded operation, padded with loads of highly paid staff, hooked up to all of these informational data based systems, that can be the know all and see all of everything to do with PWS. What we do have to offer comes from our volunteer members who obtain information and want to share it with others. Some things may be available in only one state but again it may be available in many. If you have found some source of help--share it with us. Give us the details that can be passed on to other members.

Another example of how you can help right now is that we have been receiving calls recently stating families have not been able to obtain medical insurance for their child with PWS. One parent stated it was costing them an additional \$200/month to cover their daughter. Please take a moment and share your information with us so we can pass it along to others.

New Brochures are Now Available

The first five in a series of brochures that PWSA is developing were completed in time for the national conference in July. They represent the hard work of several people, who deserve a great deal of appreciation. Special thanks go to Marsha Lupi, Ed.D., Dept. of Spec. Education, Hunter College, NY, and to Ralph Newbert, Ed.D., Labor Law Project Coordinator, University of Maine, for their contributions to the brochures on Education and Teacher information.

Also to be recognized are Louise Greenswag, Marge Wett, Janalee Heinemann (Committee Chair) and Al Heinemann, for contributions and review; Barbara Carter, Tenn. for cover designs and artwork; Lota Mitchell for editing; and our long-suffering laser printer, Linda Ho, in Pittsburgh for her patience and assistance in layout and design. Following is a list of available brochures:

1. *Behavior and Weight Management of PWS*...a collection of helpful tips for use by parents or others in contact with the person with PWS. (15¢ each U.S.; 20¢ Canadian/Overseas)
2. *What Parents of a Child with PWS Should Know About Education*...includes both general information for parents of any child needing special education, and specific material relating to PWS. (15¢ each U.S.; 20¢ Canadian/Overseas)
3. *What Educators Need to Know About PWS*...to give your child's teachers and other school personnel to most effectively manage the child with PWS. (25¢ each U.S.; 30¢ Canadian/Overseas)
4. *Informational (Publicity)*...an eye-catcher with information about the syndrome. (25¢ each U.S.; 30¢ Canadian/Overseas)
5. *Informational (Request for donations)*...about the syndrome and the national association, which can also be used by chapters seeking donations. (25¢ each U.S.; 30¢ Canadian/Overseas)

At Risk Families

"I am writing in response to the article 'At Risk Families' in May-June. We are an 'At Risk Family', and I think there is one more factor to add to explain why some parents may not have a plan for their child after their own deaths. We have no plan because there is no place for our 35 yr. old son to go. There is no PW group home in Oregon, and we have no relatives to take him in. Oregon is sending people out of our one state institution for the retarded, not taking them in. The thought of planning beyond our lifetime is always with us, and yes, it is painful, because we cannot make a plan for our son. One bright spot is that, through the auspices of the OR Health Sciences Ctr. Genetics Clinic, and especially Ann Hallum, a small group of parents has recently begun to meet in Oregon. One of the first items on our agenda is a group home.

Crisis Grant

"I can't begin to tell you how much it means to attend the conference. I could not have come without the CIATF grant. I see the parents of younger children and I know what they have yet to experience. I see the parents of older children and they have survived. It's been interesting to meet siblings and hear of their experiences. My daughter is not unique in her "hate" for her PW sister. The children have noticed they are not alone and that all people with PW act alike. Our daughter with PW has new little friends she wants to write to. She even got kissed on the cheek by a boy.

The conference has been super. It's great to see the dedication of these parents and professionals. I love my daughter but sometimes I resent the problems we must deal with in this syndrome. What I see is a beautiful group of concerned parents with a sense of humor. Thank you so much for your help and understanding. I'm so thankful for your organization. You really are a lifesaver."

Parents Share

"My daughter, 11, has a speech problem and is only now starting to verbalize her thoughts. It never ceases to amaze me how our food oriented children must live in a food oriented world. The other day at her doctor appointment she told me she was really hungry. I gave her one magazine which she quickly closed and handed back saying there was 'too much food inside'. I gave her a news magazine and she handed that back too. In frustration I told her to look at the paintings on the wall. She looked for a minute then said, 'Mommy there are two apple trees in that picture.' With that we both laughed at the no win situation. She is a stronger person than I am."

Another parent shared, "when my wife dropped our daughter off at a summer program, she took the aides hand as she left the room and stated, 'I hope my mother will be able to handle this'."

We're Proud!

When a Parent is Mentally Retarded, published by Paul H. Brooks Publishing Co., edited by our own board member Barbara Y. Whitman, Ph.D. and Pasquale Accardo, M.D., is now on the market. Barbara is Asst. Professor of Pediatrics and Adolescent Medicine, St. Louis University School of Medicine; Assoc. Professor, St. Louis University School of Social Services; Director of Family Services and Family Studies; KC Developmental Ctr. at Cardinal Glennon Children's Hospital, St. Louis, MO. She also shared with us a copy of "Moving Diagnosis: The Clinical Iconograph of Bumper Stickers" (also co-authored by Dr. Accardo), which was recently reproduced in *The Primal Whimper*. It's nice to know we have such talented people serving on our board.

Dear Readers and PWSA

"I hear people saying that they have everything and experienced a lot of things. Well this is true for me. I'm a live-in sitter and college student. And for the past six years I have had the experience of watching an eight year old boy grow-up. He was born with lack of muscle tone and all of the symptoms of PW, though he wasn't diagnosed with PWS until he was five years old. Now that he is eight he does a lot of things, such as swimming (he has now learned to dive) and rides a two wheel bicycle, which he learned in 15 minutes! This all helps keep his weight down. Even though, he has his days when he is not happy, he does have his good days, which I'm glad for, it makes you forget all about the temper tantrums. He gets a lot of hope, love and attention from his father, his mother and two older sisters.

I'm glad that I know him, he has helped me to learn more about him and PWS and to watch him grow has been a blessing."

Mind-Boggling Genetics and the Prader-Willi Syndrome

David H. Ledbetter, Ph.D.

There has been much press recently about "mind-boggling" new results which alter the way geneticists think about some genetic diseases. Since one of the examples cited in these brief press coverages is PWS, Marge Wett asked me to comment on what the new findings are, whether they are in fact "revolutionary," and what the role of PWS is in all of this fuss.

In classical genetics theory, as proposed by Gregor Mendel in the late 1800's and thus termed Mendelian genetics, children inherit one copy of a gene or chromosome from their mother and an equivalent gene or chromosome from their father to have two equal copies of each gene and chromosome (except the sex chromosomes, in which a female receives one X chromosome from each parent, a male receives an X from his mother and a Y chromosome from his father). Under this classical scheme, the genes from your mother and father are equal, and the effect of a disease gene would be the same whether you received it from your mother or your father.

Recent studies in mice and humans, however, suggest this simple scheme is not always true. In mice, there are many regions of the chromosomes which are now known to have different effects on development depending on whether they are inherited from mother (maternal) or father (paternal). In several impressive studies, it was shown that a particular gene was active only when it was inherited from the father; when the same gene was inherited from the mother it was completely inactive. The term "imprinting" has been coined to describe the phenomenon of differential activity of a gene depending on its maternal versus paternal origin. Imprinting of a gene is thought to occur in meiosis, the process of egg or sperm production, which is quite different in males and females.

In humans, several lines of circumstantial evidence now suggest that imprinting may affect gene expression and explain some unusual inheritance patterns. In two cases, it has been found that a child inherited two copies of chromosome 7 from their mothers and no copy of this chromosome from their father. In each case, the child had severe growth retardation, suggesting that a paternal copy of chromosome 7 is required for normal growth. This observation is quite similar to previous experiments performed in mice. A second example involves possible imprinting effects in the development of some cancers, e.g., Wilms tumor (a rare childhood kidney tumor). In this example, two mutations occur affecting both the maternal and paternal genes. However, the order of mutations appears important, in that the paternal gene is usually mutated first, followed by loss or mutation of the maternal gene.

The third example involves PWS. We have known since 1982 that about half of all patients with

PWS have a deletion of one chromosome #15, the other half of patients having apparently normal chromosomes. Merlin Butler and his co-workers showed that in almost every case, the deletion of chromosome 15 occurred in the chromosome which was inherited from the father (i.e., the father's chromosomes were normal, but a mutation occurred in one chromosome 15 during sperm formation, and this mutant or deleted 15 is the one which the child inherited). This result has now been confirmed by several other investigators. We all assumed that the reason for this bias was that deletions occurred more often in males than in females since testes and sperm are more vulnerable to environmental exposures which might cause chromosome breakage. We expected that a deletion in a maternal chromosome 15 would occur more rarely, but when it did, it would also produce PWS. This has now been shown to be incorrect, and has contributed to the "mind-boggling" new genetics.

Another syndrome, called the Angelman Syndrome (AS); described by Dr. Angelman in 1965), is associated with a characteristic face (wide mouth, prominent jaw, pointed chin), severe mental retardation with profound language deficit (can speak only a few words at most), microcephaly (small head), seizures, bouts of inappropriate laughter, and jerky "puppet-like" movements. About the only thing in common between AS and PWS is the fact that AS patients seem to have decreased pigmentation (partial albinism) similar to that observed in PWS. Classic genetics would predict that if the deletion affected the same genes on chromosome 15, all patients should have similar features. However, studies have now shown that the deletion in AS patients occurs in the chromosome 15 from the mother, in contrast to the findings in PWS. This indicates that the same deletion mutation can occur in eggs or sperm, but that the features produced by the deletion are different depending on which parent contributes it. How and why do differential "imprinting" effects occur? That's the 6 million dollar question, and that's what geneticists will not excitedly try to figure out. What are the immediate implications for PWS families and the PWSA? There are really no direct benefits regarding diagnosis or treatment of PWS individuals. However, these recent findings are certain to stimulate a substantial increase in research related to PWS, since it now appears to be an excellent model system for study of these new "mind-boggling" genetic effects. Increase research will certainly increase the awareness of PWS among geneticists, other scientists and physicians, which should also lead to an increased awareness among educators and the public. Parents and members of PWSA have always known that PWS individuals are SPECIAL; geneticists and physicians are just discovering that for themselves.

President's Message (continued)

about than to read. The first impulse on my part is to say, "what I heard was not all that bad and maybe I shouldn't bring it up." The first impulse on reading this could be, "but not in our group." Whatever the reaction the topics were brought up in discussion by the chapter presidents that attended the meeting and I was approached with the question of what to do about chapter problems engendered by such membership apathy from other chapter members.

The solution is straight forward. Become involved, express your individual responsibility as a member, vote when the opportunity occurs, be an active member of the group, no talent is too small to make a difference in the group project. Above all, remember that the reason that the PWSA and the chapters were organized is that persons with PWS need your help to survive, that they cannot do it by themselves and they are more important than our perceived differences or deficiencies. -- *Delfin J. Beltran, President*

Your Board of Directors at Work!

The PWSA Board of Directors met for nine hours during the few conference days, and some of the following actions were approved:

- A trust program will be developed for members seeking assistance in caring for their children following their death.
- Approval was given for the hiring of a part-time person in the national office for the remainder of 1989.
- The bid for the 1991 conference by the Illinois chapter was approved.
- Dr. Beltran will proceed with plans on the distribution, cost, etc. for the questionnaire information.
- Several bylaw changes were made to keep the bylaws up to changing requirements.
- The Board accepted the criteria (previously published in the GV) as acceptable usage of the restricted CIT fund. The CIT committee will formalize and publish procedures to follow to be allotted grants and loans from this fund for crisis purposes.
- One grant was approved for a project involving an exercise program for group homes serving people with PW, with the stipulation that a video tape or manual will be developed to share this information with other homes.
- Action was approved to further investigate research projects concerning PWS.
- Further meetings will be held with a volunteer member in the area of Public Relations.
- An Advisory Board Committee was formed to include Fausta and Gene Deterling as permanent members; other members shall be appointed by the Chairperson of the Board to serve limited terms.
- Approval was given to begin issuing membership cards for PWSA and to recognize patron and contributing donors in the GV.

Conference Grant

"Thank you once again for the opportunity of attending the PW conference. It is a little hard getting back into the swing of things--because one forgets the severity and consequences of this syndrome. The best part for us was meeting other parents and sharing their feelings and stories. There is such a common bond among the people and we found the atmosphere very supportive. It has certainly recharged our batteries and given us the incentive to share this syndrome with others. It is best to come out of the closet and let people know.

Also thanks to L.R. Greenswag and R.C. Alexander for the new book, *Management of Prader-Willi Syndrome*. We have purchased this book and find it very good. In our area we have about four families with small children and a couple of families with adults. Since I have come back from the conference I have contacted some parents and they seem interested in a parent support group.

Thank you so much again for all the hard work that goes on and for the continuing support us PW families need."

Our Supporters

The end of June, July, and beginning of August added \$1978.25 to our Research, CIT and Operating Funds:

RESEARCH: Frontier PW Chapter, Burke, Moss (UW), Ingalls (Lamoreaux), Uzendowski (2), Sojka, Rochester Coke (2), Masterson, Gunnison, Swartz, Hill (UW), CT Chapter, Kandall, Briggs (Johnson), and Alterman (2).

OPERATING: McAndrew, Burke, Moss (UW), Ingalls (Lamoreaux), and Uzendowski.

CIT: McAndrew, Olson (2), Bethel, Boyd, Richarz (2), Wett (Schaefer), Beltran, Goff, Horton, Haller and Rattray. Additional contributions toward the Clarissa Miller Memorial: Neill, Midwest Pub., PW MI Chapter, Boucher. To the Ken Van Swearingen Memorial: Lloyd, Sinnar, Crook, Sumner Comm., and Buckle.

We sincerely thank the above people for their continued support and also members who pay contributing and patron dues.

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 the PWSA. Duplication of this newsletter for distribution is prohibited. Quotations may be used if credit is given to PWSA. Membership dues are \$20.00 per year Individual; \$25.00 per year Family, and \$30.00 per year for Agencies/Professionals (U.S. Funds). Send dues and change of address to: PWSA, 6490 Excelsior Blvd., E-102, St. Louis Park, MN 55426. Any questions? Call us at (612) 926-1947.

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