

PRESIDENT'S MESSAGE

Is this the end of the year already? By the time this issue of The Gathered View reaches you the annual meeting of the Prader-Willi Syndrome Association will be just a few weeks away. Yes, another year has come and is going. As I write this, my 1040 is on its way to the IRS, another year has gone. This afternoon Linda and I meet with Sarah's teachers, counselors and psychologist to review her progress and plan for next year IEP, another year has gone.

Our organization is going to its third annual meeting. As I look back to the previous two, the thing I remember most clearly is leaving refreshed by the enthusiasm of the people I had talked with, ready for another year of challenge. Prader-Willi Syndrome has hit families from all walks of life and this meeting is the opportunity to share on common ground our successes and failures. We have created a format for this year's meeting that will give you the environment for communication. Let others know how you have met a problem, find out from others how they have achieved success where yours seems beyond reach. If you are new to the group, learn of the past. If you want an update on what is being done, hear about current research. Some of the committees formed last year have been active and will provide us with reports of their efforts. The annual meeting of your Board of Directors will occur and there will be an election of new members. (Please mail in your proxy from the last letter if you cannot appear in person.)

The year is passing, but another year of opportunity is coming. New ideas from the meetings will help with day to day problems that our Prader-Willi people face. Next weekend I will have the privilege of going to the Dubnoff Center in North Hollywood, California to see a new residential facility for Prader-Willi persons. In June there will also be a meeting of professionals concerned with Prader-Willi at the Children's Hospital in Los Angeles. The Minnesota group is in the final stages of getting their residential facility under way.

All of these new beginnings tell us that more and more people are becoming aware of the problems that Prader-Willi people face each day. I have recently had the opportunity to discuss Prader-Willi Syndrome with the advocate in the California Department of Developmental Disabilities. Her statement seemed to sum up a success in communication, what we want the Prader-Willi Association to achieve. Her statement was, "Six months ago I had never heard of Prader-Willi Syndrome. Now I know it to be special people with a special need that has not been

PRESIDENT'S MESSAGE, Continued

met." Hearing this from the people in government responsible for being our voice is encouraging. Reduction of budgets might speak against hope for governmental help, but it need not. The legislation has been written. It will be our job to help ourselves by being knowledgeable in the problems of Prader-Willi Syndrome and knowledgeable in the opportunities available and how to make them work. The increased need to carry out our responsibility, to be knowledgeable and to communicate can be given a boost by meeting with others and learning from others.

I hope that I will have the opportunity of seeing you at our meeting in Boca Raton. Come! Communicate! Learn!

Delfin J. Beltran, M.D.
President

VOTING

One member has asked that we clarify the voting statement on the "Proxy Ballot" you received in March. The membership votes to elect a Board of Directors. This Board of Directors votes on policies for the Association so your proxy vote is to be used by whomever you designate on the ballot as a vote for the three seats to be filled on the Board of Directors.

It is not too late to return proxies or the information sheets if you would still like to. We would like to thank the membership for the high percentage of these forms that have been returned already. We really appreciate your cooperation.

Statistics from the information survey will be included in the next GV issue.

LEGISLATIVE UPDATES

Chapter 67 - Family Health Insurance Coverage - This act amended the insurance law to include individuals who are incapable of self-sustaining employment by reason of mental illness, developmental disability or mental retardation as defined in the Mental Hygiene Law as eligible for coverage under family health insurance policies.

Public Law 94-103 - "Developmentally Disabled Assistance & Bill of Rights Act" - covers persons with developmental disabilities. The Act calls for a comprehensive state plan, using citizen advisors, handicapped persons and parents. Includes universally-affiliated training programs and the establishment of Protection & Advocacy Systems for each state.

Public Law 94-142 - "Education for All Handicapped Children Act" - Aims to have appropriate and free public education in the least restrictive environment possible. Individualized Education Plan (IEP) must be approved by parents.

Section 504 - Part of the Vocational Rehabilitation Act of 1973 is the civil rights guarantee for handicapped people. Federal agencies discriminating against handicapped citizens can have funds withdrawn. This includes educational systems, parks and recreational programs, day care centers, transportation systems and others.

PARENT SUPPORT GROUP IN CLEVELAND AREA

I am attempting to form a parent support group here in the Cleveland area, and I have been in contact with about seven families by phone within a 50-100 mile radius of Cleveland who are anxious for the group to get going. My son is being seen by a counselor at the Case-Western Reserve Mental Development Center, and he has offered me the use of their facilities for meetings. It is centrally located near the University Hospital complex. I would appreciate this being mentioned in the GV to alert other families in the Northeast Ohio area. My plan is to brainstorm at our initial meeting to formulate our various needs. Anyone wishing to join the group could contact me during the day at my office (216) 421-7340 or after 6 P.M. at my home (216) 473-0484.

Marjorie J. Henderson
5179 Edenhurst Road
Lyndhurst, Ohio 44124

GREAT BRITAIN GROUP

Last October Dr. Laurence, of The Queen Elizabeth Hospital, London, co-ordinated a meeting at which about 20 families gathered together at The Institute of Child Health (part of Great Ormond Street Hospital for Sick Children). For most of us, this was the first time that we had met with another Prader-Willi family, although most of us have been in touch with each other through the medium of several letter magazines.

At this meeting the main point that emerged was that we should form an association in this country, and try to create much more awareness of PWS. The main aims of the Prader-Willi Syndrome Parents Association will be to do this and also to give parental support.

It is possible that some British readers of The Gathered View have not yet heard from the P.W.S.P.A. in Britain. If they wish to know more about us, they may write to me at the following address and enclose a stamped, self-addressed envelope for reply.

Mrs. J. Williams
259 Whippendell Road
Watford
Herts WD1 7NN

MIDDLE ATLANTIC GROUP

The Middle Atlantic group of the PWSA, composed of PA, NJ, NY, DE, and MD members, met for their second meeting at Horsham, PA, just north of Philadelphia. The response was even better than the first meeting. More than 100 people attended, 25 of them Prader-Willi children. Again, it was the first for many.

Dr. David L. Margules, psychologist at Temple University, whose research has brought him world-wide attention, spoke on obesity. He was getting insight as to our needs as well as we were informed of his studies. Mr. Philip Kruper, psychologist at Norristown State Hospital, told of his experiences in working with PW.

The children visited the Philadelphia Zoo with numerous volunteer aides. A bus and driver were donated for their use by The Woods School, Langhorne, PA.

The next meeting is scheduled for September 26, 1981. The place is as yet undecided. Put it on your calendar now.

Doris Jane Miller

LONG ISLAND GROUP

The Long Island PW parent group met on March 23 at the Bernard Fineson Developmental Center at Howard Beach, NY. The meeting was attended by 23 members.

An informative and current presentation on PWS and the foremost problem of obesity was given by Dr. Collipp, Director of Pediatrics, Nassau County Medical Center. He is also the author of a book entitled: Childhood Obesity. He emphasized the necessity of reduction in calories and increase in the amount of daily exercise for the PW person. He explained the various endocrinological theories which might suggest the etiology for PWS. He presented slides and data charts and answered all of the questions presented.

Flyers announcing the next meeting will be mailed to anyone interested. If you are not being notified, please contact:

Mr. or Mrs. S. Levine
234 Locust Avenue
Freeport, NY 11520

SAN DIEGO UP-DATE

San Diego parents of PWS persons gathered recently to share concerns and needs. Invitations were sent parents of eleven Prader-Willi individuals and nine were able to attend. Karen Sawchenko, Program Director of Friends of Handicapped Children, was present to describe the organization and program projected for our planned PW Adult Group Home. Friends of Handicapped Children are "ready to go" as soon as start-up money is in hand. This organization comes well recommended, presently operating Care Village for twelve children plus four group homes, all dealing with those requiring assistance with behavior and other special developmental needs.

The best possible news is that the Request for Program Development Funds from the Department of Developmental Disabilities has been granted. San Diego's first residence for PW people should be an actuality by early 1982.

The San Diego parents groups is especially interested in maintaining a flow of information and will meet as often as required. San Diego Regional Center, F.H.C., PW California Foundation and the PW program at Dubnoff Center in Los Angeles will provide professional advise and assistance.

If The Gathered View reaches any parents or friends of PW's in San Diego and Imperial Counties who would like more information on our project, please contact:

Betty Shadell
4615 Skyline Lane
La Mesa, CA 92041

GROUP HOMES

It is our understanding the California group has opened their residential facility for four Prader-Willi young people, and they are presently talking about a second one.

The Minnesota group has received their final approval for their dedicated facility. Work has now begun on the rennovation of the building and all of the finishing touches required before licensing and opening.

Both the California goup and Minnesota group will be sending representatives to our conference to moderate the "Group Home Workshop". We sincerely hope, with guidance from these groups, other states will be able to follow their lead in furnishing homes badly needed in other states.

CHROMOSOMES

To the editor:

We were happy to see a summary of our Birth Defects meeting (J.T.M.) and American Society of Human Genetics Meeting (D.H.L.) presentations on chromosome studies on the PWS in the November-December issue of your newsletter. Our reports presented the first evidence for a specific cause of the PWS. Your article raised several questions about our findings that we would like to address.

In a collaborative effort, our laboratories in San Diego and Houston have performed chromosome analyses on a total of 45 individuals with a diagnosis of the PWS. Using high-resolution chromosome banding techniques, we found a very subtle chromosome abnormality in 23 of these individuals. The abnormality is a deletion, or loss, of a small piece of one chromosome number 15.

In addition to the 45 affected PW individuals, we studied 23 parents and normal siblings. None of them had chromosome abnormalities. This is not very surprising because many chromosome abnormalities are spontaneous events, occurring at random in the formation of egg or sperm cells. No other affected family members would be expected if the abnormality were the products of a spontaneous event.

During our studies on individuals with the PWS, we carried out parallel studies on 53 normal (related and unrelated) individuals in order to determine whether this subtle chromosome abnormality ever occurred in the normal population. None of these 53 individuals showed the deletion of chromosome 15. The San Diego laboratory performs high-resolution banding studies on every lymphocyte specimen it receives (mostly from mentally retarded or otherwise handicapped patients) over the last year. Of the approximately 350 individuals thus studied, a deletion in the region of chromosome 15 associated with the PWS could be proven only in patients carrying the PWS diagnosis. Approximately 50% of all PW patients have the deletion. There is no denying that the deletion if it exists in the non-PW population, is far less common than it is in PW patients. We feel that the admittedly small body of data currently available constitutes good evidence that the deletion is directly related to the syndrome (probably as a cause but we cannot rule out the possibility of its being an effect).

Twenty of the patients in our study had apparently normal chromosomes. There are several possible explanations for this. Even though we are using high-resolution techniques, it is possible that some deletions in PW individuals are too small for us to detect. Each chromosome contains hundreds of genes, and the "small" deletion we observe probably results in the loss of a large number of genes. A deletion involving fewer genes might produce the syndrome but would not be visible with a microscope. It is also possible that some of the patients had mixtures of normal and abnormal cells but because of the stringent technical requirement we imposed on our interpretation of abnormal cells, these individuals would not be recognized and would simply be scored as normal.

Another possibility is that the patients with normal chromosomes were misdiagnosed. The clinical diagnosis of the PWS is not an easy one to make, and several other syndromes can be confused with it. It may also be that the PWS is actually more than one disorder, and only some of these have a chromosomal basis. Even though a correlation between the chromosome findings and clinical features has not been completed, it is our impression that the "classic" PW cases are more likely to have chromosome abnormalities.

CHROMOSOMES (Continued)

Chromosome analysis is the only diagnostic test available for this disorder. Finding a deletion in chromosome 15 provides confirmation of the diagnosis. On the other hand, the absence of a chromosome abnormality does not rule it out. We feel that it is important for all individuals with a diagnosis of the PWS to have high-resolution chromosome analysis. In cases where a deletion is found, family studies are indicated. Even though the cases so far studied have shown no chromosome rearrangements in the parents, the possibility for such certainly exists. If both parents have normal chromosomes, the risk of having another child with the PWS is probably nil but the risk would be considerable if one of the parents were shown to have a balanced chromosome rearrangement.

We suggest that families interested in having chromosome analysis performed contact the Genetics Department at the nearest medical school. High-resolution chromosome analysis is not yet a routine procedure, and may not be available in your area. We would be happy to provide information on this procedure to your local genetics laboratory or help in arranging shipment of blood samples to a center experienced in such studies.

David H. Ledbetter, Ph.D.
Kleberg Cytogenetics Laboratory
Baylor College of Medicine
Houston, TX 77030
(713) 790-4984

J. T. Mascarello, Ph.D.
Genetics Laboratory
Children's Hospital and
Health Center
8001 Frost Street
San Diego, CA 92123
(714) 292-3443



Zonta Club donation

The Austin (MN) Area Zonta Club recently made a \$200 donation to the PWSA. Nancy Wandling, left, service project chairwoman of the Zonta Club, presented the check to Marilyn Sucha, a member of the PWSA. Zonta International has joined the United Nations in observing 1981 as the Year of the Disabled Persons. The donation by the Austin club is in keeping with this theme.

We sincerely appreciate this donation.

SUPPLEMENTAL SECURITY INCOME (SSI)

In the past couple of months we have received letters from some members that young PW adults have been refused SSI funding. The following item appeared in a recent issue of the Boulder (CO) ARC, VOICE:

Disabled persons between 18 and 21 may become eligible for supplemental security income or may have their payments increased under a law change effective last October. Under the new law, a parent's income and assets will not be considered in determining the payment, unless it is to the child's advantage.

Under the old law, the parents' income and assets were considered to belong to a disabled child who lived at home and received SSI payments, continuing until the child was 18 or 21 and still in school. This old rule still applies to children under 18.

Disabled children may qualify for payments if their parents' income and assets are low enough to meet the eligibility standards. Contact the local social security office for more information.

If any member has information regarding this funding, or has a child receiving these payments, would you please share this information with the PWSA office so we can add this information to our seeking help for these members.

TRANQUILIZERS

In the last issue of the GV we requested information regarding the use of tranquilizers in the treatment of PW patients. We received one reply with the following information:

(A psychiatrist recommended this treatment for an 18 year old whose behavior was getting progressively worse)

1 mgm Navane/4 per day really knocked everything out of order.

The first week he could hardly stay awake.

The second week he wouldn't go to sleep because he was afraid he was going to die in his sleep.

It took 12 days in the hospital to recover from this treatment.

1 mgm Stelazine

Helping to hold down cross talk, temper tantrums.

3 week treatment with no problems.

5 mgm of Valium - few apparent results.

75 mgm of Tofranil - some help.

The doctors suspect an abnormal endocrine system was the reason for low tolerance to Navane. One side benefit we notice from Stelazine is a better sleeping pattern and no more nightmares.

GENETICS

Dr. Vincent M. Riccardi, Director of the Research Cytogenetics Laboratory at Baylor College of Medicine, Houston, Texas, has offered to do free chromosome analyses on PWS patients with leukemia in themselves or in family members.



April 1980



January 1981

The April 1980 picture of Frances Robinson is her school picture. The January 1981 picture was taken on her 18th birthday. She has lost 54 pounds since June 1980, when she was positively diagnosed as having the Prader-Willi Syndrome.

The following article appeared in the Honolulu Star-Bulletin, Friday, February 13, 1981. Because the article was of great length, the Editor of The Gathered View has made the following condensed form. I do want to thank Mrs. Robinson for sharing this information with us.

Frances, who is 18 now, was in a wheelchair almost constantly. Whenever she attempted any type of physical activity it seemed she would fall and fracture a bone or two. Her mental and physical development was slow. Controlling her diet and her behavior seemed to be impossible at school. Frances' longest stay in a school system lasted a little more than a year. She is now being tutored at home.

It wasn't until last April when Frances was 17 that Mrs. Robinson discovered the problem. A letter in the Dear Abby column of the Star-Bulletin described a condition which matched Frances' perfectly, her mother said. It was Prader-Willi Syndrome. The column also gave the address of the Prader-Willi Syndrome Association in Edina, MN. After collecting information about the condition from the Association, Mrs. Robinson had the diagnosis confirmed by Dr. Brian O'Hara of Kaiser Hospital's Niu Valley Clinic.

Dr. Sharon J. Bintliff, a pediatrician and director of the Birth Defects Center at Kapiolani-Children's Hospital, said that weight control studies on Prader-Willi Syndrome patients showed that even when they take 20 percent fewer calories than a "normal" individual of the same age, height and build, the Prader-Willi patient will still gain weight. "Only by reducing their intake to 50 or 60 percent can they lose weight," Bintliff said.

"They never have a sense of satiety," O'Hara said. "So when you restrict their diet, you're not really depriving them. They are as hungry at 600 calories as they are at 3,000."

Frances Robinson, who is 4 feet 8 inches tall and weighs 104 pounds, is on a 600-calories-a-day diet. Since the Robinsons discovered last April that she had Prader-Willi Syndrome, Frances has lost 54 pounds.

HONOLULU STAR-BULLETIN (Continued)

This syndrome often goes unrecognized because many of the other symptoms point to mental retardation or developmental disability. "Ninety percent of the cases are diagnosed by parents who find out by reading of children similar to their own," said a father of a Prader-Willi child.

It often is difficult to diagnose early, Bintliff said, because "It is a diagnosis of exclusion." Usually as soon as the child "develops these crazy eating patterns," Prader-Willi is pinpointed.

But, as in Frances Robinson's case, more than 17 years can go by before the syndrome is identified. This can be very frustrating for parents, Bintliff said, especially if they don't know why their child continues to gain weight, even though they have cut back the calorie allotment. "It lays a heavy trip on the parents," she said. "They see their kids cutting down and still not losing weight. It takes a lot of dietetic counseling. And the earlier the problem is diagnosed, the better."

If the obesity can be checked early, many complications due to overweight can be avoided. Prader-Willi patients probably have a shorter life span than the average person, because their obesity leads to other problems, including cardiovascular disease.

A study reported in last November's issue of "Pediatrics", showed that an intellectual decline goes hand in hand with weight gain and advancing age of those with Prader-Willi syndrome. A weight-management program and behavior-modification techniques should be practiced, although, O'Hara said, self-control of eating habits will never be achieved by Prader-Willi patients.

"It's like a lifetime jailership, for both the parents and patient," Mrs. Robinson said.

NJ PARENT

The mother of a 17 year old son with an I.Q. of 105 would like to hear from other parents on how management of food and behavior were handled with a more independent child.

She would appreciate whatever help you could give her. Please write to the editor and we will forward a copy of your letter to her.

PROGRESS

One MO mother writes a special school for behavior modification has shown dramatic results for her 4-1/2 year old child with PW. A rehab center has also increased her gross motor skills 12 months in a 5 month period of time.

She also writes, sugar causes great hyperactivity plus stubborn and temper outbursts. Even a piece of gum is detrimental. (We have heard this comment from several parents even though a control test did not show the same reaction.)

TX PARENT

I am aware that PW come in varying degrees in the different problem areas. Our son definitely has PW but does not display many of the characteristics. I don't know if it is because he is still so young or if he may never develop some of the behavioral problems and food stealing problems.

Although he appears to be a perfectionist and has "fixed ideas", this child has a sunny outgoing personality and makes friends with everyone he meets. He is also very affectionate. We have to watch his food intake but he is most cooperative.

My son's teacher (Specal Ed) tells me he appears to be very bright and picks up what the older children in his class are learning. With his background, however, he will be screened at age 5 or 6 for learning disabilities, and at present, gets physical and occupational therapy.

I would be interested in knowing how other PW persons are reacting/did react at age 4 and at what age it was learned they had this syndrome. Our son was three.

Our geneticist has told us not to look for all the characteristics of PW in the future, but it is still distressing to see. I hope other parents of younger children will write the GV editor.

(Several doctors have told PW parents the same as this mother's geneticist. Certainly it is helpful to understand about the likelihood of weight and/or behavior problems, but every child is an individual. We also have to have confidence that research is going to play an important part in the lives of young children with PW.)

PA PARENT

In October we had an emergency situation with our 27 year old PW son that we would like to share with others with the hope that the knowledge we gained from this near-tragedy might be helpful to them.

J is a resident at a state hospital in Pennsylvania. Because of his need to eat apart from others, he does not go to the cafeteria. While eating lunch, he choked on a piece of liver. Fortunately, a nurse was close enough to see what was happening and she immediately tried the Heimlich Maneuver. Due to J's obesity, she was not successful. She then called for emergency help which is available within minutes. By this time J lost consciousness.

When the physician arrived, after much effort, he was able to open a small airway. J was in intensive care and required the help of life sustaining devices. He developed pneumonia and due to poor response to medication and a persistant low-grade temperature, it was necessary to do a bronchoscopic procedure to aspirate the material which entered the lungs during the time they tried to resuscitate him. It was during this procedure that it was discovered that J had an under-developed trachea — the size of a small child's. J remained in the hospital for 10 days and recovered completely. Psychological testing and other evaluations were done at the hospital and there was no evidence of further brain damage.

We feel that the difficulty with the Heimlich Maneuver and the discovery of any underdeveloped trachea are concerns that PW parents and professionals should be aware.

HANDBOOK "PRADER-WILLI SYNDROME - A HANDBOOK FOR PARENTS" by Shirley Neason. \$2.50 first copy to members; \$3.50 additional copies and non-member cost.

OVERVIEW "AN OVERVIEW OF THE PRADER-WILLI SYNDROME" by Lota Mitchell, M.S.W. \$2.50 for members; \$3.00 non-member cost.

BOOK "PRADER-WILLI SYNDROME" by Dr. V. Holm, Dr. S. Sulzbacher, and P. Pipes, M.P.H. A collection of papers presented at a Washington Medical conference. \$24.50 U.S.; \$30.50 U.S. Funds, Airmail Overseas.

CONFERENCE PAPERS

1979 National Conference Papers, PWSA. Complete set \$10.00

Papers include:

Gene Deterling, Conference Introductions

Hans Zellweger, M.D., PWS

Betty Schultze, Ed.D., Synopsis of Speech & Language Testing, One Child

Margo Thornley, Study of 8 PWS people in a residential home

Shirley Neason, Adults with PWS

Bryan Hall, M.D., Diagnostic & Counseling Dilemmas

Eleanor Watson, R.D., Residential School Program for 2 PWS pre-teens

Questions during and at close of conference

1980 National Conference Papers, PWSA. Complete set \$10.00.

Papers include:

Gene Deterling, Conference Introductions

Andree Walczak, M.D., Case study of larger group of PWS children

Patty Otto, Abstract of Sucrose Behavior Study

Betty Schultze, Ed.D., Due Process in Education report

Behavior Change Contract Ideas

Bryan Hall, M.D., Features (less frequent?) of PWS

Stacy Roback, M.D., Treatment of Undescended Testes

Questions at close of conference

PWS BOOK DELAYED

The publisher promised us a delivery of the book by Dr. Holm, et al the early part of April. I was assured this was a definite date but upon not receiving it, a call to the publisher resulted in a promise of delivery for mid-May.

Please forgive the delay. I will ship the book as soon as it is received. I have placed an order for 200 copies and have already received orders for 100 of these. I do not know if I will be able to obtain any more at the same price or not, so if you want one at this price, please order now.

Mail order and make check payable to:

PRADER-WILLI SYNDROME ASSOCIATION
5515 Malibu Drive
Edina, MN 55436

Number of copies ordered: _____

Cost: \$23.00 plus \$1.50 packaging and postage: TOTAL COST: \$24.50 U.S. FUNDS
Overseas airmail - TOTAL COST: \$30.50 U.S. Funds

Include address if different from label on reverse side.

CANADIAN MEMBERS - Banks will not honor the statement, "Pay in U.S. Funds" on personal checks. These checks will be discounted. We have allowed for this discount on our membership fee but have not on our PWS book orders. Rates must be checked if you are using a personal check.

1981 NATIONAL PRADER-WILLI CONFERENCE - BOCA RATON, JUNE 25, 26, 27

We are sorry the registration forms were inadvertently delayed but we hope with the early release of the date you were able to plan ahead and save these dates for the conference.

A chance to talk in a workshop setting, as well as informally, will be a very important part of this conference. We hope many of you will bring your PW children and young adults so that they too will get the same opportunity.

Time is running out for registration. We hope you all have sent it in by now.

A SPECIAL THANK YOU

The Senior High Resource Room Class at Bloomington Kennedy High School, Bloomington, Minnesota have been collating, folding and labeling The Gathered View the last six months. One of our members' PW child is a part of this class. A VERY SPECIAL THANK YOU is extended to each and everyone of the class for a job well done.

THE GATHERED VIEW is the official newsletter of the Prader-Willi Syndrome Association and is sent to all members. Membership dues are \$10.00 per year for U.S. members; \$15.00 per year for Canada, Mexico and overseas members. Send dues and change of address notices to PRADER-WILLI SYNDROME ASSOCIATION, 5515 Malibu Drive, Edina, MN 55436.
