The Gathered View

National Newsletter of the Prader-Willi Syndrome Association

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Chromosome 15: The Link to Prader-Willi

by Suzanne B. Cassidy, M.D.

What Is This Molecular Genetics Test About, Anyway?

Many of you have heard, in one context or another, about the new molecular genetics testing that applies to PWS. It is all very new, and usually quite confusing. It is also very hard to know whether this is a test that is appropriate for your family, and how to go about getting it. The following summary of what the test is about, and some guidelines about who are the appropriate persons or families to be studied.

First, it is important to understand the chromosome findings in PWS.

Recent information indicates that about 70% of people with clinically typical PWS are missing a small piece of chromosome material from the middle of one member of the chromosome 15 pair; called a deletion. All of our chromosomes come in pairs, and normally we get one member of each pair from each parent. For those who have PWS with a deletion, it is the member of the chromosome 15 pair which was received from the father that is missing a piece of material. Whatever genetic information is normally found in that missing material, its absence seems to cause the features that we

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Prader-Willi Syndrome Assoc.
6490 Excelsior Blvd. E102
St. Louis Park MN 55426-4797
800-926-4797 612-926-1947
Fax 612-928-9133

Office Hours: 8:00 am - 4:30pm CST
Tere A. Schaefer, Executive Director

Spotlight on Committees

THE LONG-RANGE PLANNING COMMITTEE

Three years ago, the Board (under the direction of the Long Range Planning Committee chaired by D.J. Miller), set four priority goal areas. PWSA has since focused on these four areas: Education (about PWS), Crisis Intervention, Fund Raising, and Development of the National Office. This year the new Long-Range Planning Committee (chaired by Frank Moss of California) will be working on developing a formal long-range plan for PWSA, to be based upon newly developed priorities.

This is an exciting and important project, and Frank is well qualified to lead it. He has extensive business experience, first working for others, and now in his own business. He is also the father of Melissa, age 19, who has PWS. Other committee members include Stewart Maurer (Ind.), Bud Bush (Cal.), Paul Alterman (Ga.), Jim Kane (Md.), and Tere Schaefer, Executive Director.

The committee's first task is to conduct a survey, being designed now, of the membership to discover the ideas, desires, and goals that they would like to see reflected in such a plan for their Association. The input of all will be vital in the Committee's work of planning for PWSA's future.

Keeping You Informed

In June, the membership of PWSA received a letter from its Board of Directors regarding a proposed new organization, the ``National Prader-Willi Foundation.'' At the 1991 national conference in July, it was determined that the two organizations (PWSA and the proposed Foundation) are and will be totally independent of each other.

PWSA neither endorses nor seeks to obstruct Mr. Tarakan's efforts.

(cont. from page 1)
associate with PWS. A small percentage of the individuals who have PWS but no deletion have some other chromosome change that involves the same part of chromosome 15 that is deleted in patients who have a deletion. The remaining 25% of people with typical PWS have recently been found to have a newly detectable alteration of the chromosomes called disomy. Actually, the chromosome 15s are normal in those people, but instead of having inherited one chromosome 15 from the father and one the mother, both chromosome 15s have come from the mother and there is no chromosome 15 present from the father. The end result is similar to the deletion: there is an absence of the father's contribution to the important portion of chromosome 15 in regards to PWS. It is only the mechanism that produces the deficiency in the father's contribution that distinguishes the two groups of patients. There are no obvious clinical differences between those with disomy, except that more people who have the deletion tend to be fair in coloring.

We don't yet know exactly what causes disomy. However, we believe that some people who have PWS with disomy started out having three copies of chromosome 15 (1 chromosome from Dad, and 2 from Mom), and then lost the father's chromosome. Alternatively, it could have been that there was no chromosome 15 in the sperm and by coincidence there were two chromosome 15s in the egg, and so the embryo survived. With only one chromosome 15, an embryo could not survive.

We don't really need to know the mechanism that causes disomy in order to see the relevance of disomy for PWS. First, it explains why some people don't have a deletion, but still have PWS. Second, it confirms that this is a condition with a very low likelihood of happening more than once in a family (low recurrence risk). There has never been a family who had a child with PWS with a deletion who had a second affected child. So far, among the few dozen individuals who have been found to have disomy, there has also never been more than one affected child. In the very few families in the world who have had more than one child with PWS, there has been neither a deletion nor disomy in those who have been studied.

There have been a few people who have some but not all of the symptoms of PWS, who are called ``atypical,'' and who do not have either deletion or disomy. These might be misdiagnoses. It might also be that there is considerable overlap between PWS and some other disorder which is caused in a different way. A third possibility is that these people have a deletion that is too small to be seen by chromosome analysis, but could be found if molecular testing was done.

Testing can determine whether the child with PWS has a deltion, disomy, or neither.

Molecular testing is a process which is very different from that of chromosome analysis.

In chromosome analysis, we are essentially using powerful microscopes and special staining techniques to look inside the cell at the nucleus where the chromosomes are found. Molecular genetics uses

a technique of biochemically dissecting the DNA. DNA is a very long molecule that makes up the whole length of each chromosome and contains within it information for making all the proteins of the body. Individual genes are just sections of that DNA molecule. Molecular testing looks at smaller or larger sections of DNA using pieces of DNA that are complementary to certain parts of the chromosome. These complementary pieces of DNA, called probes, are usually labeled in some way (with radioactivity, for example) so it is possible to measure whether a particular part of the DNA is present or not. In looking for deletions (either those visible with chromosome analysis or those too small to be seen by that technique), one merely looks to see whether there are some pieces of the DNA material missing. To look for disomy, however, it is necessary to see whether there are pieces of DNA from both parents or only from one parent. Therefore, a blood sample is needed from both parents and the affected individual in order to compare the DNA characteristics. These characteristics are just normal variations between individuals which allow the scientist to distinguish among different people's DNA. In many cases, it is possible to distinguish between the DNA of the mother and the father, and therefore tell which one or ones went into the child who is affected with PWS. Occasionally, the two parents have DNAs which are sufficiently similar by the testing process that is available today that we are unable to distinguish between them. Such families are called ``uniformative.'' This means that we are unable to tell whether or not there is a deletion or disomy using the current technology available. New probes are developed all the time, and it may well be that other probes would be able to distinguish between these parents in the future. In such cases, repeat testing at a later date may be indicated in order to determine whether the child has a deletion, disomy, or neither.

There are four major reasons to perform molecular testing in a family who has a child with PWS.

- When the child is not "typical'' (``atypical'') or when the diagnosis is uncertain for whatever reason, it would be possible to confirm the diagnosis or eliminate it by doing molecular testing.
- In some families, it is important to have an objective test which confirms the diagnosis in order for the parents to feel comfortable that this is really the reason for their child's problems. Such cases could also be tested.
- It could be argued that molecular testing would be of value to determine the likelihood that a couple might have a second affected child. We know that there are only a few families around the world who have had more than one affected child. We also know that, in those cases described after 1981, none of the affected children in such families had a deletion. A few such families have been studied now that we can test for disomy, and none have had disomy, either. However, most of them have not yet been tested. We assume that the risk of having a second child affected with PWS when the first child has a deletion is very low, certainly less than 1%. It is estimated that in families that have a child with

disomy the risk is less than 2%, although again we really do not know; it could be much lower. If neither deletion nor disomy is present, then we really do not know what the recurrence risk is, and in such families this information may be helpful in family planning.

■ There are some people who have been identified as having PWS because a deletion of 15q was found, even though the individual does not look like they have PWS clinically. Such cases are ideal for the testing, since the chromosome analysis is not 100% reliable.

Our knowledge and understanding about molecular genetic testing and its relation to PWS are very new. Over the next few years, many pieces of information will be clarified. Most likely, molecular testing for PWS will become routinely available through commercial genetics labs, much like testing for a few other genetic disorders has already become available.

If you feel that this testing is appropriate for your family and wish additional information, please contact PWSA or myself.

Suzanne B. Cassidy, M.D.
Associate Professor of Pediatrics
Section of Genetics/Dysmorphology
University of AZ College of Medicine
Tucson, AZ 85728
(602) 795-5675

President's Message

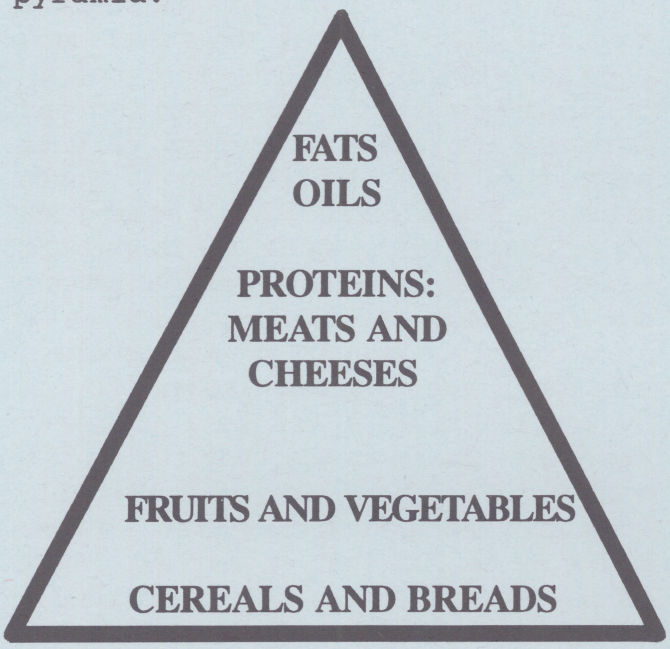
The Prader-Willi syndrome conference in Chicago was the tenth national conference Al and I have attended. It's hard to describe the warm sense of community we felt there. It still feels exceptionally good to meet new families and greet old ones. Where else can you go:...Where no one gets upset when someone's child walks into the middle of a conference session crying and hollering for Mom?...Where a parent doesn't have to feel mortified because their ``child'' walks into the restaurant and starts eating the cakes and pies on display?...Where everyone is delighted to see the young children and tries to handle their young parents with care and sensitivity?...Where everyone feels a warm glow of seeing our young people with PWS of all shapes and sizes dressed up for the dance and meeting their dates?

Wonderful education and sharing sessions aside, those adults who stopped by the dance will all tell you that these are the memories that remain the most vivid to them. There was Michael, who can't hear and is heavy set, but who had a smile from ear to ear all night as he did his Jimi Hendrix imitation with his ``air guitar'' and danced to the music by feeling the vibrations from the floor. And then there was debonair Curtis who charmed all the girls and a few of us old women by telling us how beautiful we looked. Curtis also acknowledged he knew he looked great in his suit and tie. The Don Juan award though, probably goes to Timmy; who attended the conference for the first time, but managed to get three dates for the dance! (His mother gets the ``Most Embarrassed Parent'' award for when all three girls showed up in the lobby at the same time to meet him!) Weak muscle tone and weight aside, our young people can usually out dance us

(Pres. Message continued on pg 7)

Lite Fast Foods...Are they really better?

There are many questions about basic nutrition most of us do not know; unless there is a medical reason for being aware, such as having a child with PWS.
If your knowledge is not as sharp as it needs to be, here is a very simplified guide. Remember the pyramid:



Each level indicates how much you need from each group each day.
It is easy to control the intake from each of the groups with a planned menu and dieting at home. However, in our ``on the go'' society, meals are often eaten out. What are some good strategies for eating out? It is the generally believed that the ``lite'' menu is the best menu. But are ``lite'' meals really better? Looking at the chart below one can see the ``lite'' menu items often contain far more fat than is needed by anyone, dieting or not!

Food	*Cal.*	*Fat (g)*	*Percent Cals. from fat*
McDonald's			
McLean Deluxe	320	10	28.13%
Filet-O-Fish	440	26	53.18%
Burger King			
BK Chicken	379	13	30.87%
Double Whopper	935	61	58.72%
Wendy's			
Grilled Chicken	340	13	34.41%
Big Classic	570	33	52.11%
KFC			
Lite n'crispy 2 drumsticks	242	14	52.07%
Chicken Sand.	482	27	50.41%
Hardee's			
Oat Bran Muffin	440	18	36.82%
Real Lean Delux	577	25	38.37%
Big Delux	730	41	50.55%

What are the alternatives then, if fast food is the choice? One could reduce intake tomorrow for extra calories and fats consumed today; order simply the smallest item on the menu (single burger) or a kiddie meal; take away the bun or the breading off the chicken; or make a deal with your child and let him or her eat only half of the meal (if that's possible!).
There is also hope for future better labeling of foods. New guidelines for standardizing nutrition labels may be implemented by the year 1995. Maybe some day we won't have to be a algebra majors to be able to figure out the fat content of the foods we buy.

("President's Message Continued ")

parents. One young couple with PWS solved their problem of fatigue and weight by putting their chairs facing each other, and sat leaning forward with their arms wrapped around each other, swaying to the music. One mother told me her son and another young woman with PWS don't communicate all year and don't talk at the conference--but have a standing date for the banquet and dance. Each year Phil and I also have a standing ``date'' for one dance. This year he was getting more than a little worried because I was late getting there, and smiled with relief when I finally showed up. When Phil and I stepped onto the dance floor, he started our dance with the usual question...''What did you have to eat at your banquet?''

The Chicago Chapter did an outstanding job of pulling together the largest PWS conference in our 13-year history of conferences. With 408 adults and 241 in the YAAP program, of which 161 were people of all ages with PWS, it was a challenge to say the least! There were many excellent presenters, yet adequate time was left for personal sharing. And, of course, we had them rolling in the aisles with our ``Laughter After Tears'' panel. (It's not that we're that funny, but that people really needed that release.) The sale of educational materials was at a record high, and there were exciting reports from across the nation and Canada about tremendous outreach that is taking place as more and more people learn about the syndrome. It was great to see two of our board members, **D.J. Miller** and **Stewart Maurer**, being honored at the banquet. They may not be as ``flashy'' as some, but have always been there when needed (and D.J. came through by offering to help host the national conference in Philadelphia next year). They have always worked for the good of the syndrome, and are part of the backbone of the organization.

With growth and change also come losses. We announced at the conference that **Marge Wett** has resigned as executive director. This association would not be what it is without Marge, and we could never repay her in time or money for what she has done for us. Fortunately, she is not ``abandoning ship,'' and will remain on in a part-time secretarial role; thus will be available for consultation. Also to our good fortune, Tere Schaefer, who has been assistant director for ten months, has agreed to take the position. (She has been in training as a sibling all her life.) Marge certainly has a right to work less hard, and with less hassles. In a few months, when I call the national office, I hope to hear that Marge is on vacation in the Caribbean!

I hope you can all ``cruise'' to the conference in Philadelphia next year.

Janalee Tomaseski-Heinemann, President PWSA

Mark your Calendars
1992
Philadelphia, PA

14th Annual PWSA Conference
Thursday, July 16
through Saturday, July 19

Pre-Conference Day
Wednesday, July 15

IEP: A Team Process

Remember the parent is a Very Important Part of the team!

Individualized Education Programs (IEP's) are an important part of the education process. The plans are developed through a team process. Members of the team may include: teachers, the school principle, school nurses, special aides, and (without a doubt) the parent(s).

There are five things you can expect the plan to include:

1-What the child has accomplished to date. Levels of performance evaluated may include intellectual functioning, academic functioning, communicative status, motor ability, sensory status, health/physical status, emotional/social development, behavior skills, functional skills, and vocational or secondary transition training.

2-What they are striving to accomplish by the end of the plan. Whether the plan will be an initial, annual, or interim plan.

3-Services which will be provided for the child. Including whether they will take place in a regular classroom or resource room, and in a regular public school or a specialized school.

4-Who will be responsible for the child during <u>all</u> times of the day.

5-Evaluation process for the plan and when it will take place.

As a parent, there are also six important areas that you should go into an IEP meeting prepared to address:

1- Relevant information about the child's skills, interests, strengths, and weaknesses.

2- Special medical concerns that everyone involved with the child should know. Make sure everyone understands any technical terms. Collect materials overtime which support the child's needs. In this way, everyone has the same knowledge and expectations.

3- All school activities, including non-academic areas such as lunch and recess. Also include other areas such as art, music, and special adaptive physical education.

4- Any and all questions you may have concerning your child's education. It helps to have them written out in advance, that way you can be assured all your concerns will be addressed.

5- Make sure everything needed is <u>specifically written</u> into the plan. General statements such as ''we'll work on this'' do not outline a plan of action by which evaluations can be done. Three months from now anything not written into the plan will be forgotten.

7-Finally, be prepared to evaluate the plan and make sure it corresponds to your knowledge of the child's abilities, yet also provides challenges.

PWSA has brochures designed for both teachers and parents regarding the education process; let us know if you need any.

Persistant * Persistant * Persistant*

B e
E nergetic

S elf-confident
U nderstanding
C onvincing
C ooperative
E nthusiastic
S pecific
S uccinct
F ocused
U nique
L ively

I nformed
N atural

O bservant
B elievable
T enacious
A ware
I nnovative
N imble
I nstinctive
N egotiating
G enuine

S incere
E fficient
R ight
V igorous
I ndustrious
C ute
E ducated
S pirited

Optimistic*Adamant*Unwavering

Optimistic*Adamant*Unwavering

Persistant * Persistant * Persistant*

Discouraged Mom Enlightened

Remember when you first found out about the syndrome? And the way you felt when someone actually gave you a name for what might possibly be wrong with your child ? Here is a trip down memory lane for those of us who have been through this.

Recently my son was diagnosed with PWS. I am a single parent of two children, aged five and three. At my PW son's birth he was immediately placed in intensive care for what now is obvious reasons. He was in two hospitals in Mississippi, and no one there could tell us for sure what was causing these problems. They told us that he would probably never walk or talk, and most likely would be a ``vegetable'' if he even survived.

> My doctor now says PW children are six times the work and ten times the reward.

The doctor's consoling words were, ``You can have other children, the likelihood of this occurring again is slim.'' As I kept this in mind, every accomplishment he makes, every hurdle that he overcomes, is a blessing. He is the sweetest, most loving child I've ever met. He, now at three, is very concerned for other people and instantly loves everyone he meets. People whom I have never met before will stop us wherever we go and talk with my son and they will know his name. It continues to amaze me how such a small person with so much love in his heart can stir the hearts of so many people. They don't soon forget it. Recently, one of his doctors here, who has contact with other children with PWS, made the statement that these children are six times the work and ten times the reward. The reward is far greater than that. He is a very special person. I want to encourage him to strive for the best that life has to offer and help him to realize those things for himself. I think that my son has great potential and it would be a terrible injustice on my part for me not to do all I can to help him grow to be a person that he is proud of.

Ten Years and Still Going

Oakwood residence, a designated PW group home with 15 residents in Minneapolis, celebrated ten years since opening their doors. The celebration included an Open house with neighbors and friends, Bingo playing, Bean Bag tossing, and picking from the fish pond. The grand finale included a raffle of an array of lovely prizes. Some staff members celebrated not only the home being open a decade, but their personal eighth or nineth year of employment. Most remarkably, there have been only two changes in residents in all this time.

Congratulations Oakwood 9/1/81- 9/1/91

Your Support Means So Much

(From Mid-June thru Mid-September, our various funds increased due to your support)

Research and Operating: $3110
CIT: $291
Memorials: $1400

CIT: Olson(4), Beltran(2), Parent

Research & Operating: Weiner, Chappell, Margolis, Gemini Inc. (Wett), UW-Inland Valley, PW of GA, Vanderveer Grp (Herman), UW- Ventura (Moss), UW Monroe(Vermuelen), Millipore Fd(Gravallese), Alterman, Reisenman

Memorials: Diamond: Stone(2), Agardi, Schmults Kanish: Bryan, Burg, Mains, Reynolds Schweitzer:Padesky,Kircher, Meyer, Otte, Becker, Dirck, Poelvoorde, Andrews, VandeMoortel Padesky (Bishop),Sojka (memorials), Carlson(Tucci), Gordon(Mondel), Levine (Leonard), Echols (Echols), Uzendowski (Trusz & Costellano), Gordon (Loupus), Charton (Mansourian), Dorsey& 1st Presb. Church (Simmons), PWSA CT (Mayer), Gilbert (Ziifle)

Contributors:(Dues $40-$99) Tarcia, Jackson, Seal, Huffman, Fuller, Eager, Breneisen, Carroll, Werner, Stege, Foley, Bunker, Bartlett, Haller, Smith, Spears, Brock, Couture, Wassell, Lundh, Stickle, Ranberg, Buchanan, McHargue, Hilton, Levine, Huibregtse, Minos, Paolini, Krauer, Conry, Moss, Rosetta, Simmons, Drummond.

Patrons: (Dues $100+) Kirchhoff, Draffen, Neason, Ayotte, Nanzig, Shadell, LaBossiere, Holm, Deterling, Quadrel, McManus, Masterson, Thomas, Dooley, Trimble, Howkinson, Kircheff, Atwood, Hruska, Gulling, McAndrew, Wissmann.

Two contributions, totaling $1500, were also received for the specific purpose of purchasing new equipment for the office.

We wish to thank all of the above contributors and also many members for the supportive messages that have been conveyed to us in recent months.

PWSA Surveys Workforce

Many thanks again to all who participated in the studies of occupational and living placements. The results were presented at the National Conference in Chicago. Briefly, here are some of the highlights of the workforce study:

A Total of 173 surveys were returned
87 Males, 86 Females
Average Ht.-59.39 inches Average Wt.-153.6 lbs.
Is the current work placement a proper placement:
137-Yes 36-No
Where are the placements:
118 in Sheltered Workshops
16 in Community Employment
28 in Day Activity programs
7 in Occupational Training
4 were Unknown
Is Food control needed: 153-Yes 20-No
How is food controlled:
56 Limited Supervision, 76 Constant supervision, 16 have no access to food, 5 were unknown

Behavior: Solutions for Behavior problems ranged from behavior modification techniques to positive attention by staff or token economy systems.

Specific tasks included: Folding laundry, packaging, veterinary hospital assistant, assembly/collating, landscaping, janitorial, library page, nurses aide in nursing home, warehouse stocking.

Many parents ask: "My child will be graduating from High School next year, what can I be looking at for them to be doing with their days?"
A case manager/social worker may be of assistance in exploring options with you. Many issues will arise: transportation, amount of time to be worked, and the need for food control. Also of concern is the amount of pay that the individual will receive. If a young adult with PW earns more than $500 per month, they will be considered ineligible for other benefits such as SSI and Medicaid.

Options to explore:
Sheltered Workshops--Supported Workshops employ only those with disabilities. Disabilities range in both areas of mental and physical capabilities. Most workshops receive jobs ranging from simple production (putting poker chips into sleeves) to complex production work (gluing parts together). Many workshops also offer what are called enclaves or mobile crews. Enclaves are workers trained in a small group and supervised together in an ordinary work setting. Mobile crews are teams which move from business to business (such as grounds keepers or janitors).

Community (Supported) Employment-- Work in a competitive environment, often accompanied by an on-the-job coach. The job coach remains working with the individual until the task has been mastered and the employer is comfortable with the placement. Once this occurs, the job coach continues to periodically check on the continued success of the placement.

Day Placements-- These facilities offers a wide variety of activities. Activities include vocational training, arts and crafts, basic living skills, physical education, other educational classes, and sometimes basic assembly tasks.

Each of these placements offers advantages and disadvantages. Try whichever you believe will be the best opportunity for your young adult. Finding job coaches or staff people who will believe and listen is one of the most important aspects of finding an acceptable placement.

Placement Study Results

Sixy one individulas from across the country responded to the questionnaire on living placement. Those surveyed were parents/guardians who have children with PW over the age of 21. Of those who responded, 45 are placed outside of their parent's home, and 16 were not placed. In evaluating those placements: 37 felt their children were in a good setting, and 24 did

not feel they were in a good placment. Of those who did not believe their child was in a proper placement, 97% of them were in placements other than a designated PW group home. Placements of inidividuals included: 21 in designated PW homes, 14 in Mixed group homes, 6 in state facilities, 2 in Independent living, 3 at home, and 15 unknown. Average number of residents in a group home was 5; and the typical staff ratio was one staff person for every three residents.

This brief study still leaves many questions about what is a best and/or proper placement for adults with PW. However, it does tend to support the feeling that there is a great need for more designated group homes.

Many state chapters are in the process of attempting to meet this challenge. National is also in process of developing video tapes to aid those working on developing group homes. The most difficult part of developing these guides is that each state, and often each county within a state, has different rules and regulations concerning the opening of a group home. The old adage "it isn't what you know, but who you know," definitely holds true. It is also a matter of knowing how to play the game of politics. The parents in Missouri also learned that the media can be most helpful in making others aware of the critical need for assistance. One thing is for certain, a group parents working together is the most effective way of getting action.

The Gathered View is the official newsletter of the Prader-Willi Syndrome Association and is sent to all members. The opinions expressed in The Gathered View represent those of the authors of the articles published, and do not necessarily reflect the opinion or position of the officers and Board of Directors of PWSA. Duplication of this newsletter for publication is prohibited. Quotations may be used upon credit given to PWSA. Membership dues are $20 for an individual, $25 per family, $30 per agency/professional. Any dues change of address or letters to: 6490 Excelsior Blvd. E102, St. Louis Park, MN 55426-4797. Questions or comments regarding this publication or PWS call 800-926-4797 or 612-926-1947 or Fax 612-928-9133.

Prader - Willi Syndrome Association
6490 Excelsior Blvd. E102
St. Louis Park MN 55426-4797

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