PWSA of WI, Inc.

Prader-Willi Syndrome Association of Wisconsin, Inc

A Heartfelt Message

In many ways, it seems like just yesterday when I walked into the exam room at Riley Children's Hospital and was told my son had Prader-Willi syndrome (PWS). I remember being stunned that after 4 long years of testing we had finally received a diagnosis; however, within seconds the fear kicked in as my mother, a nurse, started to cry. Unbeknownst to me, she had been surfing the Internet throughout the years searching for a diagnosis and during her search she had read many articles about PWS. The majority of the articles had highlighted every negative aspect about PWS and had made the future for anyone who was diagnosed with the disorder seem bleak. As she started talking about the various articles I remember thinking, "What had I done to deserve this? What kind of life is my son going to have? What can I do to help my son have the most successful life possible?". In addition, I felt extreme sadness over all the things my son and I would never experience due to his disorder….no sports, school parties and no birthday bashes with cake and ice cream. Well I am ecstatic to say…boy was I wrong.

My son is now a 6'2", 183 pound 19 year old who is healthy and happy. Although he has faced some struggles throughout his life, he has also experienced many successes. He has had the pleasure of participating in Special Olympics swimming, baseball, basketball and bowling, along with horseback riding through a local organization. In addition, he has been able to participate in many of the special events at schools such as music programs, field trips, holiday parties and most importantly **GRADUATION**!! Oh, and did I mention he also has gotten to have birthday bashes with cake and ice cream? Although modifications do need to be made at times, the reality is individuals with PWS can live happy fulfilling lives!

I know right now, you may feel numb and be experiencing a wide variety of emotions. Please keep in mind that you are never alone. As president of PWSA of WI, Inc., I can honestly say that any questions or feelings you may have – have been asked or felt by fellow parents before. There is always someone a phone call or email away. Our state office is located in the home of Joshua Escher, a former teacher and uncle to a niece with special needs. He answers our toll-free phone and responds to emails promptly. If he doesn't know the answer to your question, he will do his best to get it. PWSA of WI, Inc. has become a strong voice for persons with PWS as well as those who support them.

The world today is a different place than it was 19 years ago. And the world tomorrow is filled with hope and new answers to old questions. We continue to learn so much that is helping all who are affected by this disorder. There is a lot to learn but don't worry – take things one-step at a time. We are here to help you.

Most important – enjoy your child. First and foremost, you have a beautiful, loving child that will allow you to appreciate and celebrate so many things in life that we take for granted. Do what you would do for any of your children – sing, play, laugh and … love him/her. Treasure the childhood moments and remember always that your child is a child first.

Whenever you are ready, please contact our office. You will soon discover that you have new friends and other parents that are here to help and guide you if you ever want or need it. You are beginning the role of being a special parent. PWSA of WI, Inc is here to support and help you.

Sincerely,

Crystal L. Boser
Parent and President
PWSA of WI, Inc.

38 S Main Street #226 • Oconomowoc, WI 53066
920-733-3077 Email: ProgDir@pwsaofwi.org
Website: www.pwsaofwi.org

Dear Parent or Caregiver:

I would like to take the opportunity to introduce myself. My name is Amber Gaulke and I am a proud parent of a 4 year old son Greysen who has Prader Willi Syndrome. My husband, Andrew Gaulke and I have two daughters as well ages 7 and 15 and currently live in Pewaukee WI. In my free time I enjoy the outdoors, camping, fishing, and watching my kid's sports. I am also a board member of the Prader-Willi Syndrome Association of WI, Inc.

As a parent of a young child with Prader Willi Syndrome, I understand just how difficult it can be to receive the diagnosis of PWS. Faced with many challenges of this magnitude is overwhelming, to say the least. You may find yourself like me asking a million question, searching the Internet for answers. Although the Internet is a wonderful place to find information, I believe it is also very important to connect on a personal level with other families with children with PWS.

PWSA of WI would like to connect with families affected by Prader-Willi Syndrome, in particular those with children ages 0-5. There are a few ways we can connect families with young children, but to do this we are asking for your help.

One way to connect is through the closed Facebook Group named PWSA of WI Young Families 0-9 Chat. It is a great group for families to meet and exchange information with other families experiencing similar challenges. It is especially beneficial for families of younger children to meet and learn from families who have many of the same experiences and much knowledge to share. After you join, please introduce yourself and post a picture of your child…we all love the adorable babies and children!

If you are not a member of Facebook, please take the opportunity to join. Then for those who are not already members of the group, search PWSA of WI Young Families 0-9 Chat and request to join. Please email topics you would like to see on the page and any questions regarding the Facebook group to me at a.gaulke@yahoo.com.

We would also encourage you to fill out the enclosed releases and return. One is a release form granting or denying usage of any photos taken of your child. The second form is a release form for sharing contact information. By releasing this information, I would be able to share your contact information with other parents of young children, in hopes of connecting families to email, chat, etc.

We would like to let you know that the Prader-Willi Syndrome Association of WI has a web site that is updated regularly with information about PWS and events. You can access the website at: www.pwsaofwi.org. While you are on the website, please take the time to apply online for a FREE membership.

Please don't hesitate to contact me with any questions you may have. I look forward to talking with you.

Thanks You! Amber Gaulke

PWSA of WI, Inc.

a.gaulke@yahoo.com

262-844-4921

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Words of Wisdom for the Newly Diagnosed

Lota Mitchell M.S.W

Well, it's happened to you. That which you thought only happened to "other people". You have a "special needs" child. You have a child who has Prader-Willi syndrome (PWS).

Maybe it is the baby you've been waiting for and dreaming about for nine months, or a toddler for whom you have finally gotten a diagnosis. It may even be an older child. But whatever the age, you will no doubt find yourself grieving for the child you didn't have, that beautiful, normal child of your dreams.

What comes first is denial: "No! It can't be. It can't be happening to me and my child and my family." And maybe you're angry because it **is** happening to you and your child and your family. Maybe you find yourself hating the doctor who gave you the bad news, or hating other parents whose kids are just fine, or even hating your baby or child. You may feel despair and fear as you try to face reality, and there's a long, long road ahead to the destination of acceptance.

It's important that you don't deny your feelings. They are there; they exist; and they are real. If you discover that you are hating the child you love so much, then the next step may be hating yourself. The anger and fear and despair that threatens to overwhelm you needs a safe place to be expressed, no matter how negative those feelings are. Often when a person confronts, expresses and deals with such feelings, that person may be more able to cope, not only with the feelings but also with the demands that inevitably come with a child with PWS.

So who for you can provide that safe place? It should be a person whom you can trust to listen without judgment, to hear you out when you need to talk, and to keep in confidence that which you are saying. For you, would that be a member of the family? A beloved friend? A minister, priest or rabbi? Parent support groups also can provide that kind of support, plus the assurance that you are not alone. Even the Internet with its email discussion lists can be helpful. In some cases, you might want to seek counseling professional.

Now there's more to consider in this situation than just you and your little one. There's your marriage. Your husband or wife is just as important as he or she was before this happened. A marriage is like a green plant – watered regularly, it will thrive and grown, but without attention it will wither and die, or at the very least, wilt. Have dates; express your affection; comfort each other; do things together separate from the child or children.

This is often a time of great stress for marriages, especially when the spouses do their grieving differently and at different rates. Sometimes the husband will bury himself in his work and be unavailable both emotionally and physically to his wife who want to talk about it all the time. Or the wife may be so focused on the child's needs that she no longer had any time for her husband. So if you find that your marriage is drooping, make haste to get some counseling. You're already unhappy – don't increase it.

There are also your other children, if you have them. Life shouldn't revolve around the child with PWS. Yes there will be demands on our time, your strength and your spirit, but they need you too. And they can be a source of comfort and joy, if you let them. Yes, even in the midst of sibling squabbles, broken toys and runny noses!

Take some time out to enjoy the rest of your family and other facets of your life. Do you like play bridge or tennis? Do you want to sing in the church choir? How about the book review group you've been invited to join? Or a get-away weekend with your spouse? Do it! Fit that interest in somewhere so you don't lose yourself as a person. Don't be afraid to use respite care, be it babysitters, professionals, grandparents, or other relatives or friends. Teach them (what is needed), try them (to be sure they can meet those needs) and then trust them (to do it).

As your child grows, be sure to provide discipline. Remember that the word "discipline" does not mean punishment but teaching – teaching your child what is appropriate and what is not appropriate; teaching your child what is expected of him or her. Because your child has PWS doesn't mean that he or she should be pitied and not given the discipline you would give any other child. You want your son or daughter to have self-esteem and to become as acceptable part of society as possible and consistent discipline with realistic expectations is a route to those highly desirable ends.

You may not feel like it right now, but you are going to be able to laugh again…

Be sure too, to notice your child's strengths – what he or she can do. After all, there are individuals with PWS who have made Eagle Scout, been bar mitzvahed, won awards for horseback riding, performed in dance recitals, been in movies, participated in community service, spoke at training sessions for agencies serving persons with PWS, won bowling tournaments, made the Honor Society, fasted to raise awareness of hunger in poor countries …who have brought not just problems but joy to their parents. Granted, no one with this syndrome has been signed to a professional football team but elected to public office or written a best-seller but who knows what can happen?

Last but not least is laughter. Sometimes if we don't laugh, we cry. You may not feel it right now, but you going to be able to laugh again. Cultivate that ability! Your child is going to say and do some funny things as he or she grows. Enjoy these moments, write them down and get a good chuckle out of the humor. After all, being the parent of a child with PWS means trying to figure out how to lock up the apple tree!

WISCONSIN Prader-Willi SYNDROME ASSOCIATION (USA) SAVING AND TRANSFORMING LIVES

QUESTIONS & ANSWERS On Prader-Willi Syndrome

Q: What is Prader-Willi syndrome (PWS)?

A: PWS is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that can lead to excessive eating and life-threatening obesity.

Q: Is PWS inherited?

A: Most cases of PWS are attributed to a spontaneous genetic error that occurs at or near the time of conception for unknown reasons and therefore is not inherited. In a very small percentage of cases (2 percent or less), a genetic defect that does not affect the parent can be passed on to the child, and in these families more than one child may be affected. A PWS-like disorder can also be acquired after birth if the hypothalamus in the brain is damaged through injury or surgery.

Q: How common is PWS?

A: It is estimated that one in 12,000 to 15,000 people has PWS. Although considered a "rare" disorder, Prader-Willi syndrome is one of the most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified. PWS is found in people of both sexes and all races.

Q: How is PWS diagnosed?

A: Suspicion of the diagnosis is first assessed clinically, then confirmed by specialized genetic testing on a blood sample. Formal diagnostic criteria for the clinical recognition of PWS have been published (Holm et al, Pediatrics 91, 398, 1993) and further refined (Gunay-Aygun M, Schwartz S, Heeger S, O'Riordan MA, Cassidy SB. The changing purpose of Prader-Willi syndrome clinical diagnostic criteria and proposed revised criteria. Pediatrics, 108(5):E92, 2001). The diagnostic criteria and also laboratory testing guidelines for PWS can be found on the PWSA(USA) website www.pwsausa.org. as well as in the 4th Edition of "Management of Prader-Willi Syndrome" textbook. Genetic testing is now very accurate for the diagnosis.

Q: What is known about the genetic abnormality?

A: Basically, the occurrence of PWS is due to lack of several genes on one of an individual's two chromosome 15s - the one normally contributed by the father. In the majority of cases, there is a deletion - the critical genes are somehow lost from the chromosome (deletion). In most of the remaining cases, the entire chromosome from the father is missing and there are instead two chromosome 15s from the mother (uniparental disomy). The critical paternal genes lacking in people with PWS have a role in the regulation of appetite. This is an area of active research in a number of laboratories around the world, since understanding this defect may be very helpful not only to those with PWS but to understanding obesity in otherwise normal people.

Q: What causes the appetite and obesity problems in PWS?

A: People with PWS have a flaw in the hypothalamus part of their brain, which normally registers feelings of hunger and satiety. While the problem is not yet fully understood, it is apparent that people with this flaw never feel full; they have a continuous urge to eat that they cannot learn to control. To compound this problem, people with PWS need less food than their peers without the syndrome because their bodies have less muscle and tend to burn fewer calories.

Q: Does the overeating associated with PWS begin at birth?

A: No. In fact, newborns with PWS often cannot get enough nourishment because low muscle tone impairs their sucking ability. Many require special feeding techniques or tube feeding for several months after birth, until muscle control improves. Sometime in the following years, usually before school age, children with PWS develop an intense interest in food and can quickly gain excess weight if calories are not restricted.

Q: Do diet medications work for the appetite problems in PWS?

A: Unfortunately, no appetite suppressant has worked consistently for people with PWS. Most require an extremely low-calorie diet all their lives and must have their environments designed so that they have very limited access to food. For example, many families have to lock the kitchen or the cabinets and refrigerator. As adults, most affected individuals can control their weight best in a group home designed specifically for people with PWS, where food access can be restricted without interfering with the rights of those who don't need such restriction.

Q: What kinds of behavior problems do people with PWS have?

A: In addition to their involuntary focus on food, people with PWS tend to have obsessive/compulsive behaviors that are not related to food, such as repetitive thoughts and verbalizations, collecting and hoarding of possessions, picking at skin irritations, and a strong need for routine and predictability. Frustration or changes in plans can easily set off a loss of emotional control in someone with PWS, ranging from tears to temper tantrums to physical aggression. While psychotropic medications can help some individuals, the essential strategies for minimizing difficult behaviors in PWS are careful structuring of the person's environment and consistent use of positive behavior management and supports.

Q: Does early diagnosis help?

A: While there is no medical prevention or cure, early diagnosis of Prader-Willi syndrome gives parents time to learn about and prepare for the challenges that lie ahead and to establish family routines that will support their child's diet and behavior needs from the start. Knowing the cause of their child's developmental delays can facilitate a family's access to important early intervention services and may help program staff identify areas of specific need or risk. Additionally, a diagnosis of PWS opens the doors to a network of information and support from professionals and other families who are dealing with the syndrome.

Q: What does the future hold for people with PWS?

A: With help, people with PWS can expect to accomplish many of the things their "normal" peers do - complete school, participate in their outside areas of interest, be a productive worker under the right conditions, even move away from their family home. They do, however, need a significant amount of support from their families and from school, work and residential service providers, both to achieve these goals and to avoid obesity and the serious health consequences that accompany it. Even those with IQs in the normal range need lifelong diet supervision and protection from food availability. Although in the past many people with PWS died in adolescence or young adulthood, prevention of obesity can offer those with the syndrome the probability of living a normal life span. New medications, including psychotropic drugs and synthetic growth hormone, are already improving the quality of life for some people with PWS. Ongoing research offers the hope of new discoveries that will enable people affected by this unusual condition to live more independent lives.

Prader-Willi Syndrome Association (USA), 8588 Potter Park Drive, Suite 500, Sarasota, Florida 34238
800– 926-4797 * 941-312-0400 * Fax: 941-312-0142 * info@pwsausa.org * www.pwsausa.org

The Young Child with Prader-Willi Syndrome
Physical and Sensory Issues and Recommendations

By Janice M. Agarwal, PT, CNDT

Janice M. Agarwal, PT, CNDT is a pediatric physical therapist who is also the mother of a son with Prader-Willi syndrome (PWS). She is the author of a booklet, "*Therapeutic Interventions for the Child with PWS*" that is available for purchase from PWSA (USA). Website: www.pwsausa.org or call toll free: 1-800-926-4797.

Most children with PWS receive services from a physical therapist for many years. Today many infants and children now receive growth hormone therapy, so the degree of low muscle tone has improved. However many still face physical and sensory issues that require therapy and ongoing attention. Consult with a physical therapist for questions and recommendations about any of the strategies suggested.

Physical and Sensory Deficiencies:

All children with PWS are born with abnormal muscle tone and have physical and sensory deficiencies. As a result, it is common to see:

- Trunk/upper and lower extremity weakness
- Skeletal abnormalities – scoliosis (lateral curvature of the spine) and hip dysplasia
- Sensory integration problems or deficiencies
- Oral-motor dyspraxia - difficulty in making and coordinating precise movements, which are used in the production of spoken language, which results in severe, persisting speech production difficulties.

SENSORY INTEGRATION PROBLEMS	**What You Can Do to Help**:
Vestibular System: The vestibular system provides information on movement, gravity and changing positions (esp. head positions). **Some problems you may see include:** • Inability to use eyes and hands together in a coordinated effort. • Poor balance. Clumsiness. • Difficulty paying attention, concentrating and using reasoning • Doing the same thing over and over again. • Avoids movement or touch • Low muscle tone/hypotonia • Difficulty maintaining alertness; fatigue • Unpredictable emotions. Difficulty handling changes • Difficulty organizing self and using self-control • Poor understanding of relationships to objects in space • Poor understanding of what is said to them. (Not good at auditory processing) • Difficulty sleeping.	• Rocking, gentle bouncing, slow spinning in one direction • Rocking in rocking chair or on rocking horse. • Walking, running, hiking or swimming. • Bounce on large balls and mattresses • Rolling • Roughhousing or wrestling. • Somersaulting. • Spinning on swivel chair, "Sit and Spin" toy, scooter board or tire swing. **Calming strategies for vestibular problems** include slow, rhythmic, linear swinging or rocking, gentle, slow spinning in one direction. Gentle bouncing.
SENSORY INTEGRATION PROBLEMS **Proprioception:** Proprioception input provides an unconscious awareness of our body, its position and its relationship to other parts as well as other people and objects. It helps us know how much force is needed for muscles to contract and move. Receptors are located in all of our joints. It helps calm the nervous system. **Some problems you may see**: • Clumsiness • Exerts too much or too little pressure on objects • Tantrums – throws self on ground. • Affectionate – hugs tightly, sits on laps. Seeks enclosed/tight spaces for boundaries • Poor writing skills; difficulty coloring in lines and/or stopping activities	**What You Can Do to Help**: • Carrying heavier object – books, watering cans, suitcases • Pushing and pulling items • Crawling through tunnels/ boxes • Hanging (from monkey bars) • Jumping – on trampoline, mattress or air mattress • Pounding nails; rolling play dough • Swimming or extra bath time • "Tug of War" with blankets or ropes • Karate

SENSORY INTEGRATION PROBLEMS cont. **Tactile:** Tactile input provides us with information about light touch, pressure, vibration, temperature and pain. This feedback system helps to develop body awareness and motor planning. It has complimentary protective and discriminative abilities. Tends to detect sensory dysfunction. **WARNING ABOUT LIGHT TOUCH:** It is alerting; it may be uncomfortable. It can make a person feel threatened. **Some problems you may see:** • Not as sensitive to cuts, bruises, pain and temperature. • Does not like having teeth or hair brushed. • Drops things easily • Flicks or shakes hands, rubs face or licks/chews on lips • Picks at skin. Often does not tolerate Band-Aids. • Needs extra personal space. • Sloppy eaters and dressers • The feel of new clothes or tags in them may be intolerable. • Insects bites make some children crazy – scratch until bleeding. • Touching activities uncomfortable – finger painting, glue on hands/fingers • Don't always like tickling or petting	**What You Can Do to Help:** BRUSHING AND JOINT COMPRESSION • Provide DEEP PRESSURE/ heavy massage to the skin • Obtain a soft brush from a therapist; brush perpendicular to the arm/leg. • Start with the palm of the hand and go up the arm (like you are painting a wall up and down). • Gently but firmly compress joint in to themselves. • Go to the back. Brush up and down, side to side and horizontally. • Move to the foot and legs. • DO NOT brush – face, neck stomach, chest or genital areas. • Start with a 2 week trial. • Determine what behaviors you want to measure; see if there is improvement. • Do brushing 4-5 times per day. If you see positive changes – continue for 1 month, then reduce. • This is SHORT TERM TREATMENT – it is not forever. • May need a "tune up" after times of stress – illness, growth spurts, holidays… ROLLING • Use simple wooden rolling tool • Roll up and down back from neck to bottom. • Perform 4-5 times per day. • See improvement – continue for 1 month then reduce. • Maintain treatment daily
ORAL MOTOR PROBLEMS Eating is one of the most sensory intensive activity. Oral motor activity – sucking creates a calm, self-regulating state. It supports head, neck and trunk development. **Some problems you may see**: • Difficulty sucking or blowing through a straw. • Teeth grinding; inappropriate use of tongue or lips • Poor suck and swallow • Prefers certain types and texture of foods	**What You Can Do to Help:** • Blow bubbles, whistles, party blowers • Offer variety of food textures and contrasts: ○ *Cold or frozen foods – crushed ice chips* ○ *Warm soups/drinks, oatmeal, cream of wheat* ○ *Chewy bagels, dried fruits, fruit roll-up, gum* ○ *Crunchy pretzels, vegetables, apples* ○ *Sour/tart foods – cranberries, lemon-lime wedges, sour sprays* ○ *Sucking – use straws, lollipops* ○ *Tugging/pulling – beef/turkey jerky, licorice*
CALMING TECHNIQUES These strategies may help to relax the nervous system and reduce exaggerated responses to sensory input: • Warm or tepid bath • Deep massage, back rub, deep brushing or roller • Snuggling in sleeping bag, beanbag chair or pillow • Swinging back and forth; slow rocking, hugging • Hide out, fort or quiet corner. Reduce noise & light	**SLEEPING TECHNIQUES** • Warm bath or shower at bedtime • Use body pillows or sleeping bag • Try different types of pajamas – loose/tight, silky/cotton – see what child prefers • Swaddle infant, heavier weight blanket sleepers. • Dark blinds or shades to minimize light • Back rubs, brushing, rolling. • Clean, uncluttered room.

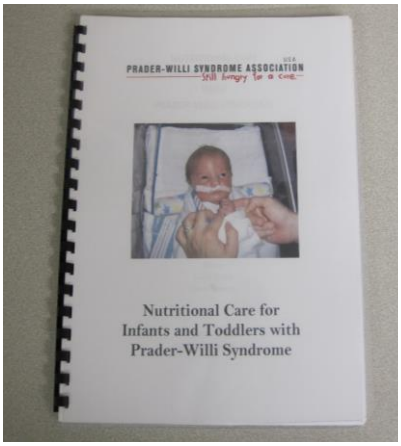
An Overview of Nutritional Resources for Parents of Infants and Children with Prader-Willi Syndrome

Becoming knowledgeable about the special nutritional needs of a child with Prader-Willi syndrome (PWS) is essential in helping him/her live a healthy life and prevent many medical problems that can be associated with overeating and obesity. Over the years, we have learned many new things about how the gastrointestinal system works in persons with PWS. This has resulted in changes in some approaches and nutritional recommendations. And … we continue to learn more.

The following are short summaries of some of the booklets available through PWSA (USA) that may be of help to those needing more knowledge and assistance in preparing and providing a healthy, low calorie diet for the child with PWS. To obtain these resources contact PWSA (USA) Website: www.pwsausa.org or call toll free: 1-800-926-4797.

Nutritional Care for Infants and Toddlers with Prader-Willi Syndrome

This resource provides information for parents of infants and toddlers (under the age of 3 years) with PWS on a wide variety of topics related to nutrition and questions about growth and development. Each chapter focuses on age-specific topics and strategies. It provides parents with information and reassurance in how to manage some of the feeding challenges – especially in those early months. It also helps families learn to address calorie, food choices and menu planning as the child ages. This booklet has excellent appendices which provide sample meal plans, menus, recipes and information about infant formulas. Also included are sample growth charts as well as additional resource information.

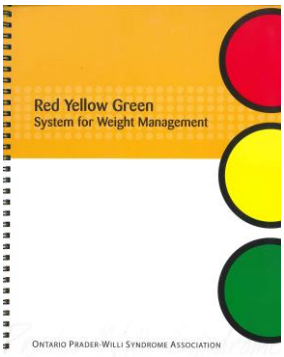


A Nutritional Guide for Parents of Children with Prader-Willi Syndrome Ages 3-9 Years

This booklet focuses on helping families of younger children ages 3-9 years with PWS gain the necessary tools and information to assist in preventing obesity and health conditions associated with it. It also helps families learn about calorie needs and food exchanges along with proper nutrition and practical suggestions on handling food-related issues. It touches briefly on food security and exercise, two areas that are an integral part of preventing obesity and maintaining good health.

Red Yellow Green System for Weight Management

Learning to meet the complex nutritional needs of a child with PWS can often seem like an overwhelming task. This booklet was specifically written for parents and family members of children with PWS. It simplifies this process by categorizing food in to three separate groups – Green, Yellow and Red. It helps families design meal plans and provides them with samples along with recipes. This booklet provides a comprehensive approach to weight management for the child with PWS including tips in developing a fitness program. This is a valuable resource for all parents and family members of a child with PWS.



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For more information, contact Prader-Willi Syndrome Association of WI Inc
Phone: Toll Free: 920-733-3077 Website: www.pwsaofwi.org

MEDICAL ALERT

A Diagnosis and Reference Guide for Physicians and Other Health Professionals

Medical Alert For Treatment Of Individuals With Prader-Willi Syndrome

Anesthesia, medication reactions

People with PWS may have unusual reactions to standard dosages of medications and anesthetic agents. Use extreme caution in giving medications that may cause sedation: prolonged and exaggerated responses have been reported. Water intoxication has occurred in relation to use of certain medications with antidiuretic effects, as well as from excess fluid intake alone.

High pain threshold

Lack of typical pain signals is common and may mask the presence of infection or injury. Someone with PWS may not complain of pain until infection is severe or may have difficulty localizing pain. Parent/caregiver reports of subtle changes in condition or behavior should be investigated for medical cause.

Respiratory concerns

Individuals with PWS may be at increased risk for respiratory difficulties. Hypotonia, weak chest muscles, and sleep apnea are among possible complicating factors. Anyone with significant snoring, regardless of age, should have a medical evaluation for obstructive sleep apnea.

Lack of vomiting

Vomiting rarely occurs in those with PWS. Emetics may be ineffective, and repeated doses may cause toxicity. This characteristic is of particular concern in light of hyperphagia and the possible ingestion of uncooked, spoiled, or otherwise unhealthful food items. The presence of vomiting may signal a life-threatening illness.

Severe gastric illness

Abdominal distention or bloating, pain and vomiting may be signs of life-threatening gastric inflammation or necrosis, more common in PWS than in the general population. Rather than localized pain, there may be a general feeling of unwellness. If an individual with PWS has these symptoms, close observation is needed. An X-ray and an endoscopy with biopsy may be necessary to determine degree of the problem and possible need for emergency surgery.

Body temperature abnormalities

Idiopathic hyper- and hypothermia have been reported. Hyperthermia may occur during minor illness and in procedures requiring anesthesia. Fever may be absent despite serious infection.

Skin lesions and bruises

Because of a habit that is common in PWS, open sores caused by skin picking may be apparent. Individuals with PWS also tend to bruise easily. Appearance of such wounds and bruises may wrongly lead to suspicion of physical abuse.

Hyperphagia (excessive appetite)

Insatiable appetite may lead to life-threatening weight gain, which can be very rapid and occur even on a low-calorie diet. Individuals with PWS must be supervised at all times in all settings where food is accessible. Those who have normal weight have achieved this because of strict external control of their diet and food intake.

What Is Prader-Willi Syndrome?

- A disorder of chromosome 15
- Prevalence: 1 : 12,000-15,000(both sexes, all races)
- Major characteristics: hypotonia, hypogonadism, hyperphagia, cognitive impairment, difficult behaviors
- Major medical concern: morbid obesity

Cause and Diagnosis of PWS

The genetic cause is loss of yet unidentified genes normally contributed by the father. This occurs from three main genetic errors: Approximately

70% of cases have a non-inherited deletion in the paternally contributed chromosome 15; approximately 25% have maternal uniparental disomy (UPD) - two maternal 15s and no paternal chromosome 15; and 2 - 5 % have an error in the "imprinting" process that renders the paternal contribution nonfunctional.

Major Clinical Findings

The following common characteristics of individuals with PWS raise suspicion of the diagnosis. Published diagnostic criteria include supportive findings and a scoring system (Holm et al, Pediatrics 91, 2, 1993).

- Neonatal and infantile central hypotonia, improving with age
- Feeding problems and poor weight gain in infancy
- Excessive or rapid weight gain between 1 and 6 years of age; central obesity in the absence of intervention
- Distinctive facial features - dolichocephaly in infants, narrow face/bifrontal diameter, almond-shaped eyes, small-appearing mouth with thin upper lip and down-turned corners of mouth
- Hypogonadism - genital hypoplasia, including undescended testes and small penis in males; delayed or incomplete gonadal maturation and delayed pubertal signs after age 16, including scant or no menses in women
- Global developmental delay before age 6; mild to moderate mental retardation or learning problems in older children
- Hyperphagia/food foraging/ohsession with food

Minor Findings

- Decreased fetal movement, infantile lethargy, weak cry
- Characteristic behavior problems - temper tantrums, violent outbursts, obsessive/ compulsive behavior; tendency to be argumentative, oppositional, rigid, manipulative, possessive, and stubborn; perseverating, stealing, lying
- Sleep disturbance or sleep apnea
- Short stature for genetic background by age 15
- Hypopigmentation - fair skin and hair compared with family
- Small hands and/or feet for height age
- Narrow hands with straight ulnar border
- Eye abnormalities (esotropia, myopia)
- Thick, viscous saliva with crusting at corners of the mouth
- Speech articulation defects
- Skin picking

Diagnostic testing

Individuals who have a number of the clinical findings should be referred for genetic testing. DNA methylation analysis confirms diagnosis of PWS. FISH and DNA techniques can identify the specific genetic cause and associated recurrence risk. (See ASHG/ACMG Report, Am J Hum Genet 58: 1085, 1996.) Patients who had negative or inconclusive tests with older techniques should be retested.

Recurrence risk

Significant only for rare cases with imprinting mutations, translocations, or inversions. All families should receive genetic counseling.

Weight And Behavior

Appetite Disorder

Hypothalamic dysfunction is thought to be the cause of the disordered appetite/satiety function characteristic of PWS. Compulsive eating and obsession with food usually begin before age 6. The urge to eat is physiological and overwhelming; it is difficult to control and requires constant vigilance.

Weight Management Challenge

Compounding the pressure of excessive appetite is a decreased calorie utilization in those with PWS (typically 1,000-1,200 kcal per day for adults), due to low muscle mass and inactivity. A balanced, low-calorie diet with vitamin and calcium supplementation is recommended. Regular weigh-ins and periodic diet review are needed. The best meal and snack plan is one the family or caregiver is able to apply routinely and consistently. Weight control depends on external food restriction and may require locking the kitchen and food storage areas. Daily exercise (at least 30 minutes) also is essential for weight control and health. To date, no medication or surgical intervention has been found that would eliminate the need for strict dieting and supervision around food. GH treatment, because it increases muscle mass and function, may allow a higher daily calorie level.

Behavior Issues

Infants and young children with PWS are typically happy and loving, and exhibit few behavior problems. Most older children and adults with PWS, however, do have difficulties with behavior regulation, manifested as difficulties with transitions and unanticipated changes. Onset of behavioral symptoms usually coincides with onset of hyperphagia (although not all problem behaviors are food- related), and difficulties peak in adolescence or early adulthood. Daily routines and structure, firm rules and limits, "time out," and positive rew,ards work best for behavior management. Psychotropic medications, particularly serotonin reuptake inhibitors such as fluoxetine and sertroline, are beneficial in treating obsessive-compulsive (OCD) symptoms, perseveration and mood swings. Depression in adults is not uncommon. Psychotic episodes occur rarely.

Developmental Concerns

Motor Skills

Motor milestones are typically delayed one to two years; although hypotonia improves, deficits in strength, coordination, balance and motor planning may continue. Physical and occupational therapies help promote skill development and proper function. Foot orthoses may be needed. Growth hormone treatment, by increasing muscle mass, may improve motor skills. Exercise and sports activities should be enrouraged and adaptations made, as needed. Proficiency with jigsaw puzzles is frequently reported, reflecting strong visual- perceptual skills.

Oral Motor and Speech

Hypotonia may create feeding problems, poor oral-motor skills and delayed speech. The need for speech therapy should be assessed in infancy. Sign language and picture communication boards can be used to reduce frustration and aid communication. Products to increase saliva may help articulation problems. Social skills training can improve pragmatic language use. Even with delays, verbal ability often becomes an area of strength for children with PWS. In rare cases, speech is severely affected.

Cognition

IQs range from 40 to 105, with an average of 70. Those with normal IQs typically have learning disabilities. Problem areas may include attention, short-term auditory memory and abstract thinking. Common strengths include long-term memory, reading ability and receptive language. Early infant stimulation should be encouraged and the need for special education services and supports assessed in preschool and beyond.

Growth

Failure to thrive in infancy may necessitate tube feeding. Infants should be closely monitored for adequate calorie intake and appropriate weight gain. Growth hormone is typically deficient, causing short stature, lack of pubertal growth spurt and a high body fat ratio, even in those with normal weight. The need for GH therapy should be assessed in both children and adults.

Sexual Development

Sex hormone levels (testosterone and estrogen) are typically low. Cryptorchidism in male infants may require surgery. Both sexes have good response to treatment for hormone deficiencies, although side effects have been reported. Early pubic hair is common, but puberty is usually late in onset and incomplete. Although it is often assumed that individuals with PWS are infertile, several pregnancies have been confirmed. Sexually active individuals should be counseled regarding the risk of pregnancy and of genetic error in offspring (50%, except for those with PWS due to UPD). Basic sex education is important in all cases to promote good health and protect against abuse.

Other Common Concerns

- **Strabismus** - esotropia is common; requires early intervention, possibly surgery
- **Scoliosis** - can occur unusually early; may be difficult to detect without X-ray; curve may progress with GH treatment. Kyphosis is also common in teens and adults
- **Osteoporosis** - can occur much earlier than usual and may cause fractures; ensure adequate calcium, vitamin D, and weight-bearing exercise; bone density test recommended
- **Diabetes mellitus, type II** - secondary to obesity; responds well to weight loss; screen obese patients regularly
- **Other obesity-related problems** - include hypoventilation, hypertension, right-sided heart failure, stasis ulcers, cellulitis and skin problems in fat folds
- **Sleep disturbances** - hypoventilation and desaturation during sleep are common, as is daytime sleepiness; sleep apnea may develop with or without obesity; sleep studies may be needed
- **Nighttime enuresis** - common at all ages; desmopressin acetate should be used in lower than normal doses
- **Skin picking** - a common, sometimes severe habit; usually in response to an existing lesion or itch on face, arms, legs, or rectum. Best managed by ignoring behavior, treating and bandaging sores, and providing substitute activities for the hands
- **Dental problems** - may include soft tooth enamel, thick sticky saliva, poor oral hygiene, teeth grinding, and infrequently rumination. Special toothbrushes can improve hygiene. Products to increase saliva flow are helpful.

Quality of Life Issues

General health is usually good in individuals with PWS. If weight is controlled, life expectancy may be normal, and the individual's health and functioning can be maximized. The constant need for fond restriction and behavior management may be stressful for family members. PWSA (USA) can provide information and support. Family counseling may also be needed. Adolescents and adults with PWS can function well in group and supported living programs, if the necessary diet control and structured environment are provided. Employment in sheltered workshops and other highly structured and supervised settings is successful for many. Residential and vocational providers must be fully informed regarding management of PWS.

Resources For Health Care Providers

"Health Care Guidelines for Individuals with PWS" and the book Management of Prader-Willi Syndrome are available from PWSA (USA), as are other publications for professionals and parents.

Prader-Willi Syndrome Association (USA), 8588 Potter Park Drive, Suite 500, Sarasota, Florida 34238
800– 926-4797 * 941-312-0400 * Fax: 941-312-0142 * info@pwsausa.org * www.pwsausa.org

A MEDICAL REFERENCE GUIDE

For Parents and Caregivers

What Is Prader-Willi Syndrome?

A disorder of chromosome 15

Occurrence in the Population: 1:12,000 to 1:15,000 (both sexes, all races)

Major Characteristics: low muscle tone, poor growth in early infancy, small external sexual organs, excessive eating developing in early childhood which could lead to obesity if not externally controlled, learning difficulties and difficult behaviors

Major Medical Concern:
Extreme obesity and consequences relate to over-eating.

Cause and Diagnosis of PWS

Prader-Willi syndrome (PWS) is caused by a loss of genes on chromosome 15 normally contributed by the father (paternal). This can occur in three ways: (1) approximately 70% of PWS cases have a deletion of part of the long arm (15q11-q13 region) of chromosome 15, thus missing these genes on the paternal side of chromosome 15; (2) approximately 25% have maternal uniparental disomy (UPD), meaning two chromosome 15s from the mother (maternal) and no paternal chromosome 15; and (3) 2-5% have an error in the "imprinting" process that makes these genes from the paternal chromosome 15 to be present but nonfunctional.

PWS is not inherited, but occurs by chance, and there is no known cause. The reoccurrence of PWS in the same family happens only in rare cases of the "imprinting" error referred to above. All children in whom PWS is suspected should have genetic testing, which is very accurate using current techniques. If diagnosis is confirmed, it is recommended that the family receive genetic counseling.

Weight and Behavior

Weight

Individuals with PWS have excessive appetite due to failure of the brain to tell them when they are full. Overeating and obsession with food usually begin before age 6. It is important to understand that this urge to eat is an actual physical disorder, overwhelming, difficult to control and requiring almost constant supervision.

Individuals with PWS require fewer calories due to their low muscle tone, low metabolism and inactivity. To create the proper calorie diet, seek out a registered nutritionist / dietitian who is familiar with the syndrome or willing to learn. Regular weigh-ins and a periodic diet review are needed. The best meal and snack plan is one that the family and caregiver are able to apply routinely and consistently. Weight control depends on restricting food from the individual with PWS and may require locking the kitchen and food storage areas. Daily exercise (at least 30 minutes a day) is a must for weight control and health. To date, no medication or surgeries have been found that would eliminate the need for strict dieting and supervision around food. Growth hormone treatment, because it increases muscle mass, may allow for a higher daily calorie intake and a better quality of life.

Behavior

Infants and young children with PWS are usually happy and loving, showing few behavioral problems. Most older children and adults, however, do have difficulties with behavior control, often happening when going from one activity to the next or with unanticipated changes. These behavioral problems usually begin at about the same time as the obsession with food, though not all behavioral problems are food related. Behavioral difficulties usually peak in adolescence or early adulthood. Daily routines and structure, fi rm rules and limits, "time out," and positive rewards work best for behavior management.

Developmental Concerns

Physical Development

Average developmental milestones are typically delayed one or two years. Although the low muscle tone improves, problems with strength, coordination and balance may continue. Physical therapy and

occupational therapy help to promote muscle development and improve coordination. Foot and/or ankle supports may be needed, Growth hormone treatment, which increases muscle mass, may improve muscle development. Exercise and sports activities should be encouraged.

Speech Development

Low muscle tone may create feeding problems and delayed speech. Speech therapy should be started in infancy to help with feeding issues and speech development. Sign language and picture communication boards help to reduce frustration and aid communication. Products to increase saliva may help with pronunciation problems. Speech development is usually delayed, and articulation problems may persist throughout life. However, speech may become an area of strength for some individuals with PWS, particularly those with UPD (both of the chromosome 15 pair from the mother).

Understanding

Individuals with PWS typically have learning problems. Areas of concern may include short attention span, inability to understand simple directions and difficulties with abstract thinking. Common strengths include long-term memory, reading ability and understanding language. Early infant stimulation is recommended; special education services and support should be started in infancy and continued through adulthood. By federal law, physical, occupational and speech therapies should be available and covered by your local school system.

Growth

Failure to thrive in infancy may require tube feeding. In order to ensure proper nutrition, infants may require special bottles or nipples (e.g., the Habermann feeder). Infants should be closely watched to make sure they get enough to eat for proper weight gain. Growth hormone levels in individuals with PWS are typically low, causing shortness, lack of puberty and increased body fat, even in those with normal weight, The need for growth hormone therapy should be considered in all individuals with PWS.

Sexual Development

Sex hormone levels (testosterone and estrogen) are typically low. Undescended testicles in boys may require HCG, testosterone, or if needed, surgery. Early pubic hair is common, but puberty is usually late and incomplete.

Other Common Concerns

Having crossed-eyes is common and requires early intervention and possibly surgery.

Curvature of the spine (scoliosis) can occur unusually early, and may be difficult to detect without x-rays. The curvature may speed up with growth hormone therapy.

Weak bones (osteoporosis) may occur much earlier than usual and may cause fractures. Provide adequate amounts of calcium, vitamin D and weight-bearing exercises. Bone density tests are recommended.

Non-insulin dependent diabetes can occur. This is usually caused by excessive obesity and will improve with weight loss. Routine screening is recommended.

Other obesity-related problems include decreased breathing, high blood pressure, right-sided heart failure, bed sores and other skin problems.

Sleep disturbances include decreased breathing with lower blood-oxygen levels during sleep, and/or daytime sleepiness. Airway obstruction may occur with or without obesity.

Skin picking is a common characteristic. It usually occurs in response to an existing sore or itch on the face, arms, legs or rectum.

Dental problems may include soft tooth enamel, thick sticky saliva, poor teeth brushing and teeth grinding.

Quality of Life Issues

General health is usually good, and life expectancy may be normal if weight is controlled. The constant need for food restriction and behavior management may be stressful for family members.

Adolescents and adults with PWS can function well in group and supported living programs if the necessary

diet control and structured environment are provided. Employment in sheltered workshops and other highly structured and supervised settings is successful for many. Residential and vocational providers must be fully informed regarding management of PWS.

Medical Alerts for Treatment of Individuals with Prader-Willi Syndrome

Negative Reactions to Medications

Individuals with PWS may have unusual reactions to standard dosages of medications. Use extreme caution in giving medications that may cause sleepiness because longer and more severe responses may occur. Water intoxication (too much body water) has occurred with the use of certain medications, as well as from drinking too much fluid.

High Pain Tolerance

Lack of typical pain signals is common and may cover up the presence of infection or injury. Someone with PWS may not complain of pain until infection is severe, and they may have a difficult time telling you where the pain is. Report any slight changes in condition or behavior to a medical professional for investigation into the cause.

Breathing Problems

Individuals with PWS may be at increased risk for possible breathing problems, such as low muscle tone, weak chest muscles and airway obstruction while sleeping. Anyone with significant snoring, regardless of age, should have a medical evaluation to look for obstructive sleep apnea.

Lack of Vomiting

Vomiting rarely occurs. Medications used to induce vomiting may not work, and repeated doses may cause poisoning. Due to the excessive eating and the possible eating of uncooked, spoiled or otherwise unhealthy food items, lack of vomiting is of particular concern. The presence of vomiting in someone with PWS may signify a life-threatening illness; therefore, if vomiting does occur, a medical professional should be contacted.

Severe Stomach Illness

Abdominal bloating, pain and vomiting may be signs of life-threatening stomach problems which are more common in individuals with PWS than in the general population. Rather than localized pain, there may be a general feeling of unwellness. If an individual with PWS has these symptoms, contact a medical professional immediately.

Body Temperature

Unexplained high or low body temperatures are common. High body temperature may occur during minor illness and in surgical procedures requiring anesthesia. Fever may be absent even with severe infection. Blood work, including a CBC (complete blood count), may be helpful in determining the severity of the illness.

Sores and Bruises

Because of the common habit of skin picking, open sores may be present and prone to skin infection. Individuals with PWS also tend to bruise easily.

Excessive Appetite

Excessive appetite and overeating may lead to life-threatening weight gain, which can be very rapid and occur even on a low calorie diet. Individuals with PWS must be supervised at all times and in all settings where food is available. Those who have normal weight have achieved this because family and/or caregivers enforce strict control of their diet.

***IN CASE OF A MEDICAL EMERGENCY PRADER-WILLI SYNDROME ASSOCIATION (USA) MAY BE CONTACTED AT 1-800-926-4797 (Between the hours of 9 am to 5 pm eastern) OR www.pwsausa.org FOR MEDICAL INFO**

Emergency Contact and Medical Information

Person's Name | Date of Birth | Sex

Parent's/Guardian's Name | Parent's/Guardian's Name

Home Phone | Work Phone | Home Phone | Work Phone

Cell Phone | Cell Phone

Address | Address

Alternative Emergency Contacts

Primary Emergency Contact | Secondary Emergency Contact

Home Phone | Work Phone | Home Phone | Work Phone

Cell Phone | Cell Phone

Address | Address

Medical Information

Medications:

Allergies:

Health Conditions:

PRADER-WILLI SYNDROME – SEE MEDICAL ALERT HANDBOOK

AT A GLANCE:

Hospital/Clinic Preference

Physician's Name / Pediatrician | Phone Number

Physician's Name / Endocrinologist | Phone Number

Physician's Name / Psychiatrist | Phone Number

Dentist's Name | Phone Number

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Insurance Information	
Primary Insurance Company	Card Number/Date
	Subscriber ID/Group Number
Secondary Insurance Company	Card Number/Date
	Subscriber ID/Group Number

I authorize all medical and surgical treatment, X-ray, laboratory, anesthesia, and other medical and/or hospital procedures as may be performed or prescribed by the attending physician and/or paramedics for the person listed and waive my right to informed consent of treatment. This waiver applies only in the event that neither parent/guardian can be reached in the case of an emergency.

Parent's/Guardian's Signature	Date

PLACE SCANS OF INSURANCE CARDS BELOW (FRONT AND BACKS)