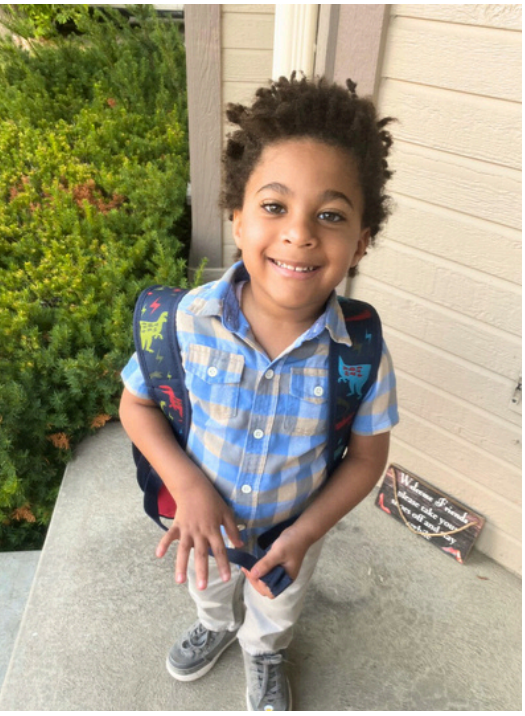
Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

Package of Hope 2



www.pwsausa.org | (941) 312-0400

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Family Support Services

PWSA | USA's Family Support team provides support to individuals diagnosed with Prader-Willi syndrome, their families, and providers. They also educate medical providers, education professionals, and professional care givers about PWS and advocate for the comprehensive needs of the entire PWS community.

What We Do

New Diagnosis Support and Parent Mentoring

PWSA | USA offers comprehensive support and education to families at the time of diagnosis and provide information about PWS. Our Parent Mentoring Program is all about making connections with other "veteran" parents.

One of the most comforting parts of this journey is talking and meeting other families. Each of our mentors has shared similar experiences. We too have been comforted and nourished by our Parent Mentor. We have wept together, and we have laughed together. We are committed to ensuring that newly diagnosed families have the most up-to-date research materials, educational literature, counseling, nutrition, and medical information all within our Package of Hope, which we have been providing to families for well over a decade. We hand select our parent mentors with each family's specific needs in mind, to guide and support you throughout your child's lifetime. The compassion and caring from these very special connections can be a lifeline of HOPE.

Diet and Weight Management

Effective weight management is a crucial part of the care of a person with Prader-Willi syndrome. PWSA | USA supports families to manage weight effectively through appropriate meal planning, environmental supports, and other effective strategies.

Behavior Support

We provide behavior support to families, school districts, and residential providers by assisting with the development of positive behavior intervention plans, review of plans, and training implementation of best practices.

Consultations and Trainings

PWSA | USA offers consultations and trainings to residential providers, school personnel, and other support staff who need guidance or increased knowledge to support their client. Consultations may be provided in person, via telephone, or teleconference.

Peer Consultations

We facilitate consultations between PWSA | USA's volunteer medical consultants and an individual's medical team, emergency room physicians, or specialty clinicians.

Insurance/Medicaid/SSI Appeal Assistance

PWSA | USA provides support letters and guidance when appealing denials for Medicaid, Insurance coverage, Supplemental Security Income (SSI) and Social Security Disability Insurance (SSDI).

Residential Placement Support

PWSA | USA has created a Residential Care Database of agencies that provide residential services to individuals diagnosed with PWS. We support families through their search for the most appropriate provider for their loved one.

Grief and Bereavement Support

Grief is the response to loss, particularly the loss of someone or something that has died to which a bond or affection was formed. Most people associate grieving with the loss of life, but grief comes in many forms. Two types of grief sometimes experienced by parents of children who have a rare genetic condition are anticipatory grief and disenfranchised grief. Anticipatory grief is just that, grief experienced in anticipation of death. Parents who worry that their child's PWS diagnosis might shorten his or her life might be experiencing anticipatory grief. Disenfranchised grief might occur when a parent mourns the loss of the child they thought they were having. PWSA | USA has a certified grief counselor on staff to help you through your journey.

If you need support, you can contact our Family Support Team by calling (941) 312-0400 or emailing info@pwsausa.org.

In partnership with state and regional Chapters, PWSA | USA is reaching more families and helping more individuals than ever before. Families turn to us for help navigating complex medical issues, support through the school years, securing safe residential care, and PWSA | USA Chapters help bring a local touch and extra support to families when they need it most.

Below you will find the contact information for every state Chapter. Please let us know if you would like someone from your state Chapter to reach out to you directly. If your state is not listed, please reach out to the Chapter in a neighboring state or contact PWSA | USA about forming a Chapter in your state or region.

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Exercise and Prader-Willi Syndrome

You have heard exercise is good for physical health, but did you know research also shows exercise is beneficial to mental health and mood? According to the Mayo Clinic, physical activity stimulates endorphins in the brain that elevate mood and encourage relaxation.

Exercise can fend off depression and anxiety in individuals with PWS and can even combat unwanted behaviors like physical aggression, self-injury, and destruction of material goods.

- If an individual has general anxiety, 60 minutes of physical activity a day is often enough to help them feel better.
- Individuals with higher levels of anxiety might need more regular periods of exercise throughout the day to feel better.
- Individuals that tend to be more explosive and aggressive might respond to regularly scheduled activities like playing basketball or running to help with emotional regulation.
- Exercise in the form of fun physical activities can also be helpful in redirecting unwanted behavior such as skin picking. If your loved one is skin picking, engage him or her in a game of catch or other physical activity which requires the use of their hands.
- Individuals with PWS and Sensory Processing issues can also benefit from exercise to help regulate their systems and decrease unwanted behaviors. The exercise routine should be based on the individual's need for sensory input. For example, if your loved one needs more physical input, like joint compression or heavy muscle work, using light weights, running, playing basketball, or performing wall pushups can help meet these needs. If your loved one needs more movement to help regulate, walking, rolling on a stability ball, or jumping on a trampoline may be more helpful.

How can you encourage someone with PWS to exercise?

- Do not call it exercise!!
- Make the activity fun.
- Do the activity with them.
- Make it part of the routine day.

- Use creative wording. Instead of, “It’s time for our walk,” try “Let us take a hike to look for birds!”
- Play tag, follow the leader, or engage in playful competition.

Exercise, while good for nearly everyone, can be a fun and interactive way to help individuals with PWS physically, mentally, emotionally, and behaviorally.

Preventing Challenging Behaviors

Behavior should not be a dirty word. Behavior is anything an organism does in response to action or stimulation. Behavior is a function of living for all of us. Good behavior does not get a lot of attention because it does not cause problems. However, when behavior causes a problem, it is labeled as bad or challenging and there is a lot of attention focused on how to fix it.

Behavior is often a means of communication. For example, when a child cries after being told that it is time to clean up her toys, she is communicating that she does not want to stop playing yet. When you see unwanted behavior occurring, approach it with a sense of curiosity and ask the questions: what is really going on here? What is my child trying to tell me? This approach helps by taking you from frustration to problem solving. You are looking at the why, rather than the what, in behavior.

Children with PWS respond exceptionally well to structure and routines. Create a routine for your family, not just your child with PWS. Your child may respond well to a visual schedule as well. Structure allows your child to know what to expect and feel safe. Build some time into your routine every day to spend some individual 1:1 time with your child doing something that is important to them. The amount of time spent doing this is not as important as your full engagement and attention. When possible, schedule this time together immediately following a less preferred activity. This accomplishes two things; it gives your child something to look forward to and it is an immediate reward for completing a task that may be challenging for them, or just one that they do not enjoy. From time to time, the routine may have to change for one reason or another. It is crucial that you preset your child when this occurs. How early to inform your child varies from child to child. For some children, too much notice of a change will increase their anxiety and cause upset. For others, the more notice and involvement in the decision the better. You know your child best.

In addition to developing structure and routine, it is important to develop appropriate expectations for your child. The expectations for rules should be consistent and your child should know what they are. Enforcing these consistently will provide your child with a sense of security like the routine and structure of the day. Developing household rules rather than rules that only apply to your child with PWS are more effective. Many children with PWS have a strong sense of fairness and will perceive rules that only

apply to them as being unfair. If the rules for your other children are different, make sure everyone in the family knows what they are and why. For example, Jonny's bedtime is 7:30 but Jane's bedtime is 8pm because Jane is older. When Jonny is older, his bedtime will be 8pm.

Know how you are going to react and be consistent in your reaction when your child does not follow the rules or meet your expectations, which will happen. If yes means yes and no sometimes means yes, especially if they keep asking, your child is not going to know what to expect. They will learn that if they keep asking you will give in. Be consistent with your expectations and your responses. Respond with firmness, validate their feelings, and provide choices whenever possible.

One of the most successful, yet underused strategies for preventing challenging behaviors is to praise your child often and sincerely. Using a 5:1 ratio of positive to negative interactions is ideal. Reinforce steps in the right direction and be specific. For example, "you did a great job putting your toys in the toy box, thank you" or "walking up the hill is really hard for you and today you walked halfway all by yourself – that was great, I'm so proud of you". Use your attention and time to reward your child's positive behavior. Praising your child not only helps to reinforce what you would like to see, it reminds you as the parent of all the wonderful attributes your child has, and the attributes certainly outweigh the challenges.

All children have challenging moments and not all behavior is related to PWS. When your child is in full melt-down mode, your reaction and response is important. After making sure everyone is safe, remain emotionally neutral and let it play itself out. Do your best to look disinterested and engaged in something else, while ensuring that they remain safe. STAY CALM. This is not the time to talk to your child or try to pull them out of the emotional state they are in. When in a full meltdown, they are unable to process any information you are giving them and talking is likely to escalate the situation. Some children may tear up books, cry, throw themselves on the floor and then fall asleep. Others may want hugs and apologize after the tantrum, do not deny them. Validate your child's feelings NOT the behavior. Do not give in to the tantrum, if you do your child will learn that tantrums are the way to get what they want.

Once your child calms, have them clean up any mess that they made. Natural consequences are most effective; your child threw their toy and it broke; they no longer have that toy to play with. Punishments are rarely effective as many children with PWS have difficulty connecting two events as cause and effect.

If you are struggling to manage your child's behavior, please call (941) 312-0400 or email info@pwsausa.org to speak with a Family Support Counselor. You are not alone; we are here to help.

Guidelines for Behavior Management

By: Kim Tula, MS

Positive behavior support is an essential tool for all caregivers of a person with Prader-Willi syndrome. It is most effective to implement positive behavior strategies early in a child's life – even before behavior challenges emerge. Many individuals with PWS feel anxious and that anxiety can lead to challenging behaviors. Managing and reducing anxiety will lead to less behaviors.

Create Consistent Routines

Consistency and routines help individuals with PWS to feel safe and secure in their environment. When they know what to expect, they feel less anxious. Using visual schedules and providing verbal prompts can be helpful. Develop a daily routine and weekly or weekend schedules that are posted.

- It is important to include meal and snack times on the schedule; reducing worry about when the next meal or snack will be.
- Include chore times, bedtime routines as well as all other activities and expectations; schedule the least preferred before the most preferred i.e., exercise/chore before lunch or dinner.
- Communicate any changes in the schedule or routine in advance.
  Note: Some children cannot handle a lot of advance notice about changes, use your best judgement, you know your child best.

Create Clear Rules

Individuals with PWS tend to be rule followers; rules help them feel safe and know what expectations are, just like consistent routines. Additionally, rules provide a guideline as to what is expected and how to behave. Once a rule is explained, it's important to practice what that rule means and looks like. For example, if the rule is "all shoes get put in the closet after you take them off," practice taking your shoes off together and then putting them away. Provide a lot of praise along the way.

- Once a rule is established, it's important to be consistent and not give in. If no sometimes means yes, your child will feel more anxious and increase their perseveration on that issue waiting for no to become yes.
- Life happens. Establish the rule that parents can change the rule if needed, however this practice should be limited.
- Encouraging your child to have a voice in the rule making, will increase their desire to follow the rules.

Create Clear Boundaries
Establish clear and consistent boundaries around expectations and teach your child what is acceptable behavior, and by default, what is not. Always say what you mean, clearly and in language that they will understand. Avoid making threats that you are unable to follow through with. For example – “put your shoes away right now or you won’t ever go to grandmas again.” Following through on what you say will increase their trust and sense of security.

- Offer preferential limited choices rather than giving positive and negative consequences. For example – “Do you want to wear your blue jacket or your red jacket?” The choices do not include, not wearing a jacket. Offering options sets clear boundaries related to what you want them to do, while giving them a sense of control.
- Avoid ambiguity and using phrases such as “maybe later,” or “we will see.” These are too vague and increase anxiety.

Be consistent! If you give into a tantrum today, tomorrow’s tantrum will be worse. By giving in to the tantrum, you are reinforcing the negative behavior that you are trying to prevent. If you have given in to a tantrum in the past, there are steps you can take to minimize the effect.

- Apologize – “I made a mistake and should not have done that, I’m sorry.”
- Explain to your child how giving in was not helpful in the past, and what to expect in the future.
- Stay strong. When working to change someone’s behavior, they will likely push harder to get you to bend the rules. Expect it to get worse before it gets better.
- It will get better!

Create a Calm Environment
Many individuals with PWS are hyper-reactive to the environment and external stimuli. The best environment is one that is calm with a calm, supportive person. Create an environment that fits your child’s specific needs.

- Limit auditory stimuli by avoiding having the radio and television on in the same room.
- Encourage the use of noise-canceling headphones in environments that you are unable to control.
- Create a safe space to go to when feeling overstimulated such as a bedroom.
- Keep sensory calming items accessible such as a swing or rocking chair, tactile items (squishy balls, brushes, vibrating stuffed animals or pillows) and calming smells such as scented lotions or essential oils on fabric.
- Keep lighting soft

Responding to a Meltdown or Tantrum
Despite your best efforts your child will have moments of upset and engage in challenging behavior. It's most effective to remain calm, matter of fact and keep a neutral affect during these situations. Some individuals are reinforced by the reaction they elicit from others during a tantrum. Paying little to no attention to unwanted behaviors will help extinguish them. Responding with compassion, while validating their feelings but not the behavior can help deescalate the situation. Remain calm on the outside, despite how you are feeling on the inside, be patient and stay firm. Once your loved one has calmed, praise them for calming. Avoid trying to talk someone out of being upset, they can't process the situation while having a tantrum or meltdown, and it will likely increase their upset.

Create Positive Consequences
Positive reinforcement is much more effective than negative reinforcement. Positive reinforcement involves providing a positive consequence to a wanted behavior, making it more likely that the behavior will occur again in the future. Praise the good, no matter how small.

Positive Behavior + Positive Reinforcement = Increased Positive Behavior

If you are struggling to manage your child's behavior, call (941) 312-0400 and ask to speak with a Family and Medical Support Counselor.

Revised 2022

Guidelines on Scoliosis Monitoring and Treatment for Children with Prader-Willi Syndrome

Author: Harold J. P. van Bosse, MD
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Background

Children with Prader-Willi Syndrome have about a 70% risk of developing scoliosis before the end of growth. Approximately 15% of children with Prader-Willi syndrome will develop severe or significant curves, requiring bracing or surgery. The earlier the curve is detected, the better the possibilities for treating the curve with casting or bracing.

There are two peak ages for scoliosis presentation in children with PWS. Under the age of four years, most of the curves begin as C-shaped, and are most likely related to the hypotonia. The second peak, centered around ten years of age, typically is the more common S-shaped curve that is seen in otherwise typically developing adolescents. Fifteen percent of curves diagnosed before four years of age will require surgical treatment, 41% of curves diagnosed after four years of age will require surgical correction, as per the 2007 PWSA-USA survey of membership. Spinal deformities in children with PWS, especially the young ones, are often picked up late, because the signs are extremely subtle on physical examination. Where most typically developing infants with even a small spine curve will have a noticeable back asymmetry, those with PWS do not. That asymmetry is caused by rotation of the spinal column, called vertebral rotation, causing one side of the rib cage to be more prominent in back when doing the forward bending test, usually the first sign of scoliosis. Children with PWS have less vertebral rotation than children without PWS with scoliosis curves of a similar size. Therefore, the child with PWS may have mild findings clinically, which are overlooked but a moderate curve radiographically. In the past, the delay in making a clinical diagnosis was blamed on obesity, but this does not appear to be the case. Fewer children with PWS now develop obesity, and often the curves are diagnosed prior to the onset of obesity. For that reason, routine screening for spine curves is important, and there should be a much lower threshold for working up clinical findings in children with PWS, compared to otherwise unaffected children.

Conditioning

Children with PWS should be encouraged to be as active as possible, particularly those activities that build up their core, strengthen their abdominals and the back muscles. In addition to sports and recreational activities, focused physical therapy and hippotherapy, emphasizing core muscle strengthening, may help improve a hypotonic

curve in a young patient with a flexible deformity. Kyphosis is the normal rounding of our upper back, which is often increased in persons with PWS. If increased kyphosis is noted to develop, the physical therapy regimen should include specific exercises for hyperextension stretching and strengthening of the upper spine. Some parents have found that their children responded well to Pilates.

Monitoring

Scoliosis in infants with PWS is unlikely to develop until the child is sitting and gravity acts across the spine. Therefore, monitoring of spine should begin when the child first sits independently, usually around the first birthday in PWS. Yearly screening radiographs of the seated or standing child should be used in addition to a clinic examination. If a curve greater than 10° is seen, spine x-rays should be taken more frequently so that any progression can be quickly appreciated.

Casting

Spinal casting has been shown to be effective if scoliosis is detected in the infant before 3 years of age, and even up until 5 or 6 years old. These body or “Mehta” casts can actually reduce the size of the curve. Curves over 20° or 25° should undergo casting, with a goal of decreasing the curves as much as possible. Many curves under 50° can be reduced to under 20° with casting, after which the child is braced for a year, with plans of being brace-free thereafter. The goal with larger curves is to control them and allow the child to grow several more years. Even curves over 90° can obtain some correction with casting, delaying surgery until after 5 years of age. The casts are changed every 2 months in children under the 2 years of age, every 3 months in children between 2 and 3 years, and every 4 months for children over 3 years of age. Casts are continued until the curve has been successfully reduced, or if correction plateaus over successive casting. The child is then braced to maintain the correction.

Bracing

Brace treatment should be considered for curves over 20° to 25° in growing children when casting is not an option. Spine braces come in different styles with different names but are all some version of a thoracic-lumbar-sacral orthosis or TLSO. For smaller, more flexible curves, a nighttime only brace is useful, trying to side-bend the spine as much as possible to straighten the curve; because of the unnatural position, these braces are not for daytime use. Larger curves, over 30°, need a daytime brace as well, countering the effects of gravity during regular daytime activities. Brace wear then increases to 22 hours per day, allowing an extra hour or two out-of-brace for physical activities. When the brace is first fitted, initial radiographs in brace should be taken to verify that the curve shows correction as compared to the out-of-brace radiograph. Smaller spine curves in children with Prader-Willi syndrome are often flexible and the goal should be to obtain a 50% correction, although this is not possible for all curves. Follow-up out-of-brace radiographs are obtained every 4 to 6 months, having the child remain out of brace overnight, until the time of the radiograph.

Surgical Intervention for Scoliosis

The goal of scoliosis treatment is to try to keep the curve under 40° to 50°, because curves larger than that will continue to progress (worsen) even after growth has ended. More importantly, severe curves at any age deform the chest cage, which can seriously affect how the lungs develop, leading to breathing problems. To prevent curve progression in adulthood, or to improve chest shape, curves that are, or will be, over 50° at the end of growth likely need surgical intervention.

Head forward alignment and junctional kyphosis

People with PWS balance themselves differently from their peers, with a characteristic head-forward position. On x-ray, this is seen as either an increased thoracic kyphosis or a cervical-thoracic (base of the neck) junction kyphosis. Experience has shown that attempting to correct the posture to “normal” can lead to failure of the surgery, as the patient works to reestablish their comfortable posture. They will lean forwards, compensating for the rigidly fixed part of the spine by bending above or below the spine rods. This can lead to a sharp kyphosis at these areas, called a proximal (upper) or distal (lower) junctional kyphosis, possibly followed by the spine pulling away from the rods. To avoid this, the proximal extent of the fixation should be as low as possible (the 3rd or 4th thoracic vertebra, called T3 or T4), and a moderate amount of existing thoracic kyphosis can be accepted.

Surgery for the growing spine

In a young, growing child, performing a definitive spinal fusion may improve the child’s deformity, but will restrict spine and chest growth. At maturity, that child will be adult sized but with a child sized chest. For these children expandable implants (rods) can control the shape of the spine while allowing for growth. In practice, I try to postpone surgery by bracing until an x-ray of the curve in its brace cannot be held below 50°. The goal of expandable implant surgery is to decrease the curve’s size initially, then prevent it from progressing while allowing for spinal growth. There are two kinds of expandable implants, one is the “growth rods” or “non-fusion spinal instrumentation” (NFSI), the other being the “vertical expandable prosthetic titanium rib” (VEPTR) device. The VEPTR device has proven to be problematic in children with PWS, primarily due to the poor bone strength of their ribs, leading to frequent failure from rib fractures. For that reason, we recommend the use of a NFSI. For PWS, the construct that has worked well is having the upper part anchored to two vertebrae in the upper spine, and the lower part anchored to two vertebrae in the lower spine, each with bilateral pedicle screws, for four pedicle screws anchor above and below. The segments are spanned with dual rods. A typical construct would be from the third thoracic vertebra (T3) to the third lumber vertebra (L3), fusing T3 and T4 to act as the upper anchor, and L2 and L3 as the lower anchor. Except in certain difficult cases, a magnetically actuated rod can be used, so that the rods can be lengthened during a regular office visit, not requiring surgery.

Magnetically actuated rods can be lengthened every 2-4 months in the clinic/office setting, whereas the manually expandable rods are usually lengthened in the operating room every 6 months, to keep up with spinal growth. Near skeletal maturity, the spine may need to undergo a definitive fusion.

Definitive spinal fusion

In the older child, a definite fusion should be performed for curves 50° or more out of brace. A curve of this size has a 95% chance of progressing, even after skeletal maturity. Curves as small as 40° may need to be fused if there is a concern of progression or poor spine balance. Whereas in idiopathic scoliosis the lower age threshold for a definitive procedure is 10-12 years of age, children with PWS often have a later growth spurt, with delayed maturation, possibly related to the use of supplemental growth hormone. For that reason, it is advantageous to wait until 12 years of age for girls and 14 years for boys prior to fusion. Bone mineral density in children with PWS is frequently low, so multi-segmental pedicle screw constructs are recommended, maximizing the number of fixation points. In the typical scoliosis seen in adolescents without PWS, the scoliosis usually has a lordo-scoliosis pattern, meaning that the upper back is actually straighter than usual when seen from the side. In children with PWS most commonly have a kypho-scoliosis, in that the rounding of the upper back as seen from the side is exaggerated. As mentioned above, care should be taken not to over correct the kyphotic deformity, as this may lead to worsening of their cervical-thoracic kyphosis. Also, as with the expandable implants, it is best to keep the upper level of the fusion no higher than T3 or T4, if possible, to prevent proximal junctional kyphosis from developing.

Surgical Planning

In children requiring anesthesia, whether for spinal casting, spine surgery with rods, or even planned interval lengthening of a NFSI, the special characteristics of children with PWS should be well understood by the entire treatment team.

Respiratory

Children with PWS have a number of respiratory issues, some related to their hypotonia, and others to obstructive and/or central sleep apnea. They are at high risk for postoperative pneumonia. The literature reflects a high rate of complications with anterior spinal procedures, and it is strongly recommended to avoid surgeries that enter the chest. Preoperatively, a consultation with a pulmonologist is required, and often a sleep study. A tonsillectomy may be indicated preoperatively for obstructive sleep apnea, and CPAP or BiPAP may be prescribed for after the operation. Postoperatively, extubation (removing the breathing tube) may need to be delayed for a few hours, or even overnight, until a patient has sufficiently awoken for a strong respiratory effort. Hypotonia may cause the patient to have a weak cough, and chest physical therapy should be aggressive.

Bone Mineral Density

Children with PWS have decreased bone mineral density and bone strength, which may lead to surgical hardware losing fixation (pulling out) or bone not fusing (pseudo arthrosis). Pre-operatively, it is important to have vitamin D and calcium levels optimized. Children with PWS who have been actively managed by their endocrinologist will likely have been on long term growth hormone and possibly sex steroid replacement. This treatment can optimize bone strength by puberty, but only if vitamin D and calcium levels have been sufficient. It is a good idea for patients to supplement their vitamin D and calcium intake, especially if surgery is being considered. Although radiographs notoriously underestimate bone mineral quality, a sure sign of an abnormality is if bowel gas pattern on a spine x-ray actually obscures the vertebra it overlaps. Another warning sign is a diet low in dairy products (milk, yogurt, cheese). A DEXA scan may be helpful if 1) there is concern that the bone mineral density is critically low, such as a history of fractures, and 2) there is enough time prior to surgery for the treatments to have an effect. In those critical cases, intravenous pamidronate and vitamin D may make a meaningful difference.

Operative planning should maximize the number of spinal fixation points for performing the instrumentation for fusion. A multi-segmental pedicle screw construct, with fixation at nearly every level has worked well. Occasionally protective bracing after surgery is needed to prevent proximal or distal junctional kyphosis.

Pain Tolerance

Children with PWS have an increased pain tolerance, which may be helpful when attempting to mobilize them after the surgery. But it may also be a reason why they awake slower from anesthesia.

Food Seeking and Gastrointestinal Issues

The family and hospital staff needs to be vigilant for food seeking behavior, and its possible life-threatening consequences. Preoperatively, the patient might violate the NPO instructions, making induction of anesthesia dangerous. Postoperatively, the children should have their diet advanced very slowly until they have normally active bowel sounds and flatus. Usually, they are hungry much earlier than other postoperative patients, but their ileus resolves a day or so later than expected. A typical protocol is 2 ounces of clear fluids every 4 hours starting immediately after surgery. If that is tolerated, it is increased to 4 ounces every 4 hours, and then gradually a soft diet is introduced. Daily abdominal radiographs are used to confirm that the ileus is improving, which may take up to a week to occur. Also, the patient's preoperative calorie restrictions should be well known by the hospital's nutrition staff and observed postoperatively. All treating staff should be aware that children with PWS do not vomit, nor lose appetite. Should either of those occur, they require an emergent nasogastric tube insertion, followed by an abdominal radiograph to evaluate for gastric dilatation,

and possibly a CT scan to rule out free air. Gastroparesis leading to gastric necrosis is unfortunately a common cause of death in children with PWS.

Skin-Picking

A constant threat to the surgical incision is the well-known characteristic of patients with PWS to skin pick, in essence scratching their wounds until they break down, leading to a dehiscence and a deep infection. This is probably the most common post-surgical PWS complication seen. Skin picking is usually direct gouging of the surgical site with the fingers, but it may take the form of rubbing their back against a wall or furniture. We use a light post-operative brace to protect the skin for the first several weeks, even for those children without a history of skin picking, as the stress of surgery may bring about new maladaptive "coping" behavior.

If the above points are carefully observed, surgery can be performed safely and uneventfully in children with PWS.

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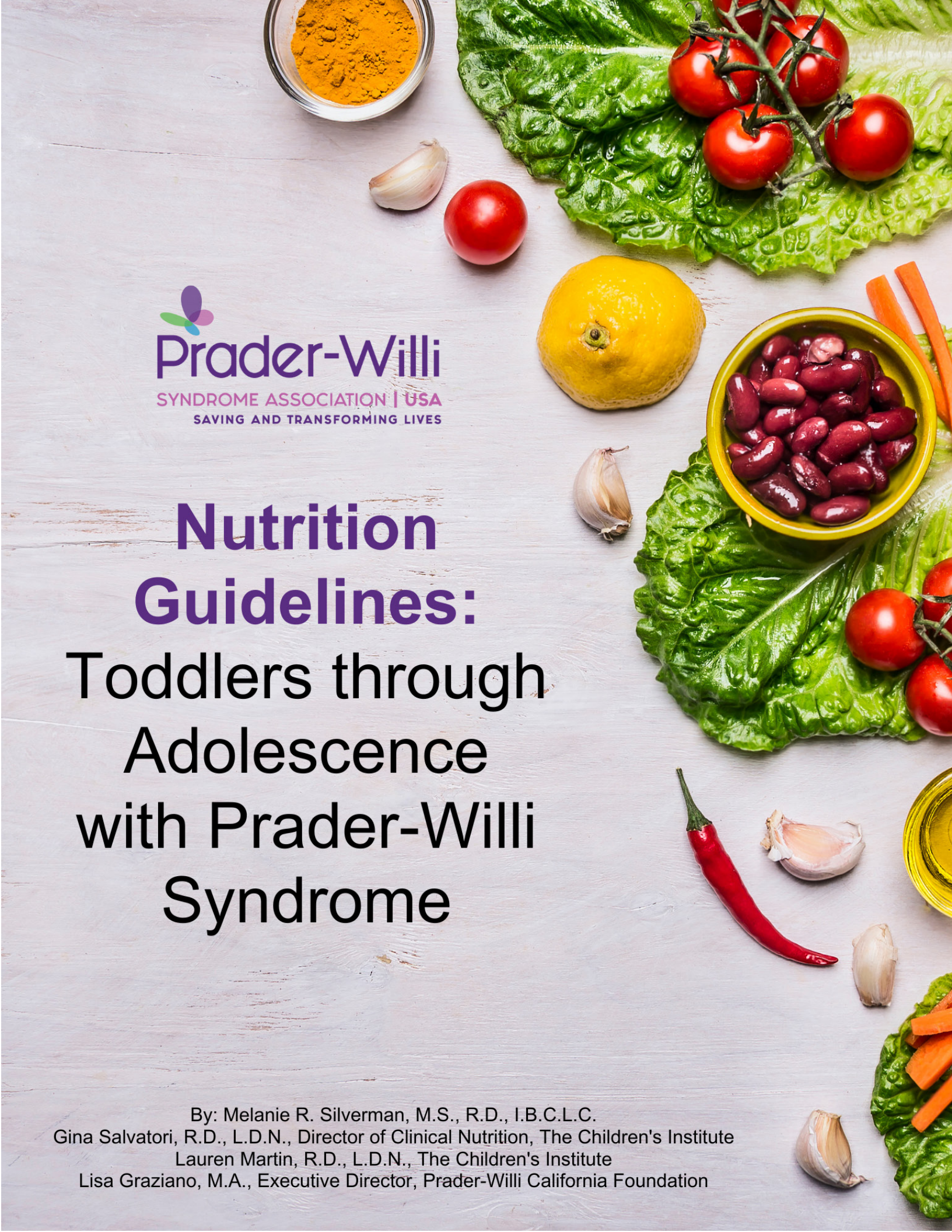

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

Nutrition Guidelines:

Toddlers through Adolescence with Prader-Willi Syndrome

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Dedication:

This booklet is dedicated to all the individuals with Prader-Willi syndrome (PWS) and their families. The authors' hope is this information provides parents and caregivers with basic nutrition knowledge they need to create the healthiest environment possible for their children with PWS. For more information on PWS, and/or state and local contacts, please visit www.pwsausa.org or contact the Prader-Willi Syndrome Association | USA directly at (941) 312-0400.

Introduction

Prader-Willi syndrome (PWS) is a genetic disorder affecting about 1 in 12,000-15,000 individuals (both sexes, all races). It has many characteristics, the most challenging being the constant obsession with food, also referred to ashyperphagia. Most individuals with PWS feel an insatiable drive to obtain food and eat most of the time. While there is not yet any prescribed medical treatment for the hyperphagia symptom, there have been successes with weight control through calorie-controlled diet, exercise, behavior management, and structured living situations. It is important to note that there is generally a spectrum among individuals with these characteristics from mild to severe.

In 2012, Miller, et.al described PWS in multiple stages:

- Phase 0: Decreased fetal movements in utero and lower birth rate
- Phase 1a: Hypotonia with difficulty feeding and may have failure to thrive (0-6 months; median age at completion 9 months)
- Phase 1b: No difficulty feeding and growing appropriately on growth curve (5-15 months; median age of onset 9 months)
- Phase 2a: Weight increasing without an increase in food or calories (20-31 months; median age of onset 2 years)
- Phase 2b: Weight increasing with an increase in food seeking (3-5 years; median age of onset 4 ½ years)
- Phase 3: Hyperphagic, food-seeking, lack of satiety (5 years-adulthood; median age of onset 8 years).
- Phase 4: Appetite for some adults is no longer insatiable and is able to feel full (adulthood)

Obesity Prevention

The prevention of obesity is one of the most important goals for the family of a child with Prader-Willi syndrome. Your child with PWS is at increased risk for obesity because the child will become hyperphagic (excessive hunger and rarely feeling full) coupled with the fact that he or she only needs about two-thirds of the calories needed by most people.

Preventing your child from becoming overweight or obese is necessary to reduce the risk of breathing problems, heart disease, hypertension, fatty liver disease and diabetes that can develop early in childhood. This can be done with a nutrient dense, calorie-controlled diet and consistent environmental controls that will help establish a healthy eating pattern for life. Obesity prevention requires a balance. The challenge is to help your child feel satisfied without giving him or her too many calories, yet provide all of the vitamins and minerals needed to grow well.

Some parents can become so fearful about their child becoming obese that they may overly restrict calories and certain food groups and then the child fails to grow. In certain situations, if weight and length have plateaued for a period of time, the degree of food restriction may need to be loosened. Parents or caregivers should monitor growth closely and speak to their physician and qualified registered dietitian for guidance on this issue.

Weight & Height Measurements

Frequent tracking of height and weight measurements are essential for monitoring your child's growth and planning an appropriate diet. Keep your own records at home. Your doctor will likely plot your child's growth measurements on a growth chart. There are growth charts specific to individuals with Prader-Willi syndrome, and the Centers for Disease Control (CDC) has growth charts for all children. The CDC charts are able to calculate a child's body mass index (BMI) to determine the degree of overweight or obesity beginning at age two years and older.

It is ideal to always weigh your child on the same scale, at the same time of the day, and in the same type of clothing (e.g. in pajamas, first thing in the morning). Measuring height measurements (marked on a wall taken with the child standing up straight) should be taken every three months. If your child is gaining weight, but is not growing taller, he or she may be getting too many calories. Check closely what he or she is eating at meals. If meals, snack and physical activity has not changed, check to see where the extra food may be coming from (e.g., refrigerator, cabinets, family members, friends, school). Ask your doctor or registered dietitian for an appropriate goal for your child's weight.

Calorie Needs

Children need calories to maintain a normal rate of growth and calorie needs change as the child grows and moves through the phases of PWS. Your child's needs may vary and in early life, a child with PWS may need a high calorie diet to grow. Once a calorie level is estimated, a child's growth should be monitored regularly to be sure that the level is accomplishing the right goal (i.e., weight maintenance or weight loss). This may require frequent weight and length/height check visits with the pediatrician to make sure your child is plotted on the growth charts according to the trends. Children on growth hormone will have significant improvement in linear growth, decrease of fat mass, and increase of lean mass. Your medical team will use your child's current intake and current weight/height or BMI to determine if extra calories are needed.

To maintain weight, children need about 20-28 calories per inch of height OR 10-11 calories per centimeter every day. A slightly overweight child's weight goal may be to maintain weight so that he or she can grow into that weight. To lose weight, children need about 18 calories per inch of height each day OR 8-9 calories per centimeter. An appropriate and safe goal for weight loss would be about two pounds per month.

Exercise

A good exercise plan is essential and important to start as early as possible. The entire family should participate, as children might need extra encouragement. Not only does this benefit the child with PWS, but it also keeps the entire family healthier. Daily walks, bike riding, swimming, gymnastics, martial arts like karate and Tae Kwon Do are examples of good physical activities. There are bound to be some hikes in your area of the country. Visit www.localhikes.com to find some.

There are also video games that involve strenuous activity like X Box games and Wii Fit. Exercise that is regularly scheduled helps burn extra calories and prevent obesity. Strenuous physical activity and/or competitive sports should be undertaken only with the approval of your child's doctor.

Nutrition Through The Years

Birth and Infancy: Many children with Prader-Willi syndrome have early feeding difficulties. At birth they appear hypotonic (floppy) and have a poor suck/swallow reflex making sucking and swallowing breast milk or formula difficult. Many children may need a nasal-gastric feeding tube or have a gastric tube placed while they gain strength to suck and swallow on their own. Breast milk is preferred, but if breast milk is not available, there are infant formulas that provide the optimal calories, protein and fat children need to grow. It's important to work closely with your physicians, dietitian and therapists to help the child gain the strength to eat by mouth and have the feeding tube, if placed, removed as soon as possible. In addition, these medical professionals can also help you with the introduction of solid foods (table foods) to your child.

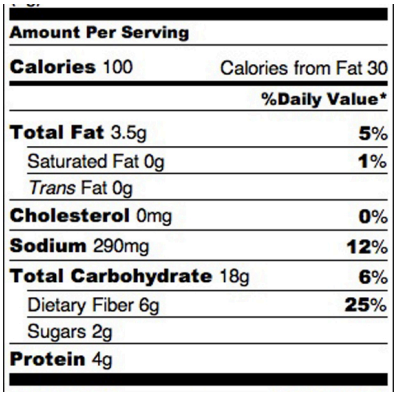
Toddlers/Preschool Age: After those early months of life, children usually begin to develop better feeding skills and are able to eat and grow appropriately without a feeding tube. Establishing good nutrition and feeding schedules is essential. Parents must pay attention to what they feed and how they feed their children. During this time, children may start Phase 2a, which means that weight will increase without an increase in calories. At this time, paying closer attention to calories and an intensive review by a registered dietitian may become crucial. If calories are reduced, a dietitian needs to make certain all nutrients are still met.

School Age Children & Teenagers: As children grow, they naturally want to be more social. This shift in their social situation will most certainly allow for more access to food. It's essential to make certain all school administrators, teachers, friends, family and coaches are aware of the diagnosis and that the child cannot have unlimited access to food. Also, physical activity is even more crucial to balance calorie intake. Scheduling at least an hour of activity 6-7 days per week is helpful to control weight.

Adults: As teenagers transition into adulthood, new challenges occur in regards to weight management and food security. Many adults participate in day programming, which promotes socialization and a level of independence but also poses an increased threat to food security. Strict monitoring of caloric intake should continue through adulthood with caloric requirements stabilizing and/or decreasing with age. Physical activity remains crucial to support ongoing weight maintenance and/or loss as well as bone health.

What to Feed Your Child

Having a child with Prader-Willi syndrome requires parents to develop a good working knowledge of nutrition. Variety is important for optimal nutrition. The nutritional goal is to offer very nutrient dense foods that are satisfying. Examples are vegetables, fruits, lean meat, chicken, turkey, fish, pork, dairy, eggs, nuts, beans, and seeds. Steer clear from foods in packages with too much sugar, unhealthy fats or salt. Reading labels is essential and ingredients lists that are full of unrecognizable ingredients are not ideal in a daily diet.



Amount Per Serving	
Calories 100	Calories from Fat 30
	%Daily Value*
Total Fat 3.5g	**5%**
Saturated Fat 0g	**1%**
Trans Fat 0g	
Cholesterol 0mg	**0%**
Sodium 290mg	**12%**
Total Carbohydrate 18g	**6%**
Dietary Fiber 6g	**25%**
Sugars 2g	
Protein 4g	

Nutrition 101…

• **Calories** are made up of carbohydrates, proteins and fats. There are 4 calories per gram of carbohydrates, 4 calories per gram of protein and 9 calories per gram of fat. Study food labels closely and review these websites for more details and information about general nutrition:
- www.kidshealth.org
- http://kidshealth.org/kid/stay_healthy/food/calorie.html
- www.cdc.gov/nutrition
- http://www.cdc.gov/nutrition/everyone/basics/

• **Carbohydrates:** This group is important for energy in the body, but there are some that are more nutritional than others that you should choose more often. The healthiest carbohydrates are fresh fruits and vegetables, whole-wheat products, brown rice, beans, peas, and lentils. Milk and yogurt are also sources of carbohydrates. Unhealthier carbohydrates are white bread, white pasta, white rice, muffins, donuts, cookies, brownies, and candy. Read food labels closely and it's ideal to look for more than 3 grams of fiber per serving for healthier carbohydrates.

• **Protein:** Protein is important for building, maintaining and replacing the tissues in the body. Muscles, organs and the immune system are made up of proteins and so it is important to have good sources of protein in the diet. Protein comes from meat, chicken, turkey, fish, eggs, dairy, beans, nuts and seeds. Each meal and snack should have a good source of protein to make for a balanced diet.

• **Fats:** Science and medicine have revised the recommendations as they relate to fat as part of a healthy diet. This recommendation also translates to people with Prader-Willi syndrome. For many years fat was considered unhealthy and low fat diets were recommended. New research has broken fats down into different components that require further explanation:

o **Monounsaturated/Unsaturated fats:** These fats have a beneficial effect on heart health when eaten in moderation and used to replace saturated fat and trans fat in a diet. Omega-3, a type of monounsaturated fat, can lower triglyceride levels and increase HDL cholesterol (the good cholesterol). Several other studies also suggest that these fatty acids may help lower blood pressure.

- Examples: Fatty fish like salmon, mackerel, herring, lake trout, sardines, and alba core tuna are high in monounsaturated fats and the very good omega-3 fatty acids. The American Heart Association recommends eating these fatty fish at least two times a week. Eating a variety of these fish will help minimize any potential effects due to environmental pollutants. Omega-3 fats can also be found in flax seeds, wal nuts, sardines, salmon, beef and soybeans. Canola oil, soybean oil, and olive oil are also good sources. While all of these foods are healthy, calories still count, so offer these foods in proper portions.

o **Saturated Fats:** These fats are found naturally occurring in some foods that provide important nutrients for a healthy diet. So the key here is to offer these foods in moderation and focus more on fats that are unsaturated as listed above.

- Examples: fatty beef, lamb, pork, poultry with skin, lard, eggs, cream, butter, cheese, and other dairy products from whole/reduced fat.

o **Trans Fatty Acids (Trans Fats):** Trans fat needs to be reduced or eliminated whenever possible. These fats can raise blood cholesterol levels and contribute to heart disease. They provide very few nutrients and are high in calories.

- Examples: These include fried foods like doughnuts, and baked goods including cakes, pie crusts, biscuits, frozen pizza, cookies, crackers, and stick margarines and other spreads.

• For the infant and very young child it is important to not overly restrict fat intake because it is still needed in the diet for adequate brain development and growth. When looking at calories, 30% of calories should come from fat.

Exchange System

The exchange system is a good plan because it is balanced, flexible, easy to follow and easy to modify as your child grows. Foods are divided into six groups based on their calories and nutrients. They are: starch/bread, vegetables, fruit, meat, dairy products, and fat. A daily food plan can be developed with the help of a registered dietitian that is based on your child's nutrient needs and food preferences. The following table, "Daily food Plan Using the Exchange system," shows the number of food choices allowed from each group for a variety of calorie levels. Serving sizes are described in the "Food Exchange Lists" at the end of this booklet.

	Calories Per Day				
	600	800	1000	1200	1400
Exchange Groups	**Number of Servings Per Day**				
Starch/Bread	2	3	3	3	3
Vegetables	7	7	7	7	7
Fruit	3	4	4	4	4
Meat	2	2	5	7	8
Dairy	1.5	2	2	2	2
Fat	1	1	1	1	1

*Fat should be used sparingly.

Standard portion sizes are too large for younger children. Foods in the correct portion size are selected each day according to the meal pattern. The foods that most children like from the exchange group are incorporated in the Food Exchange Lists at the end of this booklet.

When using the exchange system, it is still important to read food labels and compare them with the exchange list for portion size and calories. If the calories are significantly different, choose a different product or adjust the serving size.

Calorie Counting

Families may prefer to count calories rather than follow the exchange system. When counting calories, careful meal planning is necessary so that your child's diet includes all of the food groups. Careful reading of the nutrition information on the food labels is essential. Similar products may vary in calorie content. For example, one slice of diet bread has about 40 calories compared to regular bread, which has about 80 calories per slice. Be sure to serve the portion size specified in the label. Even if families opt to not count calories, it's important to have a general sense of foods that are low and high in calories when planning meals and snacks. Learning to read food labels is important and reading about nutrition is necessary.

Vitamin/ Mineral Supplements

Because the calorie level of the diet is low, it is challenging to offer all the vitamins and minerals the children need for growth on less than 1000-1200 calories per day. A complete multivitamin supplement is often recommended. One key when choosing your multivitamin is looking for the front label to include the term "complete."

One mineral that is difficult to get in adequate amounts is calcium. Calcium is important because it helps your child grow strong bones. Your child's calcium needs will change with age. One 8-oz. glass of milk has 300mg of calcium. If your child is allergic to milk, soy milk, unsweetened almond, hemp or other nondairy fortified milk can be a great option. If your child consumes too little or no dairy products, they will need a calcium supplement. For some children, limiting milk to 2 servings per day and adding a supplement at the end of the day can help save some of the calories for other foods. A registered dietitian can review your child's diet to see if they are getting the proper amounts of all vitamins and minerals.

Hydration

One of the challenges with children with Prader-Willi syndrome is their hydration status. Put simply, they often do not like to drink water so it's important to devise strategies to increase water intake. Adding the juice of oranges, lemons, or limes can help. You can also soak strawberries or apples and cinnamon overnight in water to make water more palatable to drink. Cucumbers can also flavor water. Juice and soda are not recommended because they provide excess calories that can cause weight gain.

Notes:

How to Feed Your Child

Structuring meals and snacks is essential. As a child moves from milk to more table food, setting a structure for eating provides support and predictability for your child. Posting menus and rules about mealtime behavior is also a good strategy to set expectations. But as parents, you must do it and follow through. Limit access to unsupervised buffet situations where food is readily available and easy to take.

Special Events

Treats and other high-calorie foods are a part of life. These foods, in very small amounts, may be planned into your child's diet. When your child has a treat, adjust his or her total calories by subtracting the calories from the daily total.

When incorporating treats or high-calorie foods, they must be planned ahead of time and included on your menus. Food is not to be used as a reward for good behavior. When given spontaneously, treats can be viewed by your child as a reward. Unplanned treats can also cause increased anxiety surrounding food.

Special events and holidays are important for all families. Try to make food less important during holidays. Stress decorating, gift giving, and family togetherness.

Determine what types of food will be served at your event. If limited options are going to be available, serve small meals at other points of the day and allow the individual to have a small portion of a few foods at the event. Depending on the event, packing a meal may be a good alternative.

Food Access

Along with strict diet control, your child's access to food must be controlled. Food-seeking, gorging and hoarding are all typical behaviors of children with PWS. Many families lock kitchen cupboards, refrigerators and other food storage areas. Families should have a fairly strict schedule for meals and snack times. Most children with PWS appear to like a set routine. This may help to decrease food seeking and hoarding. Serve food directly on the plates. After meals, put the food away immediately and restrict access to the kitchen.

Instruct all family members and all caregivers (e.g., extended family, teachers, neighbors, etc.) on the importance of preventing access to food. One way is not to give in to begging and temper tantrums for food. Undesirable behavior over food needs to be dealt with calmly, firmly and with consistency. Extra food is dangerous to a child with PWS. When a child's weight continues to increase and he or she is following a strict diet at home, the problem may be access to food away from home.

It is important to recognize that the degree of food issues in PWS can be a significant source of stress to the family and home environment. If the family works as a team, is consistent in approach and structure of food issues, and sibling needs are also recognized, the stress can be managed as well. Families are encouraged to seek counseling, if stresses around managing diet and food access become overwhelming.

Notes:

Prevention or control of obesity is one of the most challenging tasks for the parents of a child with PWS. It requires constant work and good communication between all who are involved in the child's care and it can be done. This is a summary of what needs to be done:

- Monitor growth closely and keep in close contact with physicians
- Balance of carbohydrate, protein and fat
- Read food labels closely
- Structure meals/snack times
- Fresh food is always best
- No juice, soda or other calorie containing drinks (unless milk is worked into diet)
- Intensive, frequent physical activity as a family
- Be an advocate for your child with PWS. Educate all parties involved in taking care of your child so they have an understanding of PWS and can keep the child safe and healthy

Food Charts

Starch/Bread (80 calories per serving) Aim for whole grains when possible.	**Serving Size**
Bread, Cereal & Pasta	
Cooked Cereals	½ cup
Ready-to-eat unsweetened cereal	¾ cup (1 oz)
Pasta (enriched or whole grain, cooked)	½ cup
Rice (white or brown), cooked	½ cup
Quinoa, cooked	1/3 cup
Couscous	1/3 cup
Granola	¼ cup
Bagel	½ bagel
English Muffin	½ muffin
Hot Dog or Hamburger Bun	½ bun
Light Hot Dog or Hamburger Bun	1
Pita (6” across)	1
Tortilla (6” across)	1
Bread (white or wheat)	1 slice
Light/Diet Bread (white or wheat)	2 slices
Dried Peas, Beans, Lentils	
Beans (cooked)	½ cup
Lentils (cooked)	1/3 cup
Starchy Vegetables	
Corn or peas	½ cup
Corn on the cob (6” long)	1
Baked Potato (small)	1 (3 oz)
Mashed Potatoes	½ cup
Winter Squash (acorn or butternut)	¾ cup
Yams or Sweet Potatoes	½ cup
Crackers/Snacks	
Animal Crackers (plain)	8
Graham Crackers (2 1/2” square)	3
Popcorn (no fat added)	3 cups
Pretzels (sticks)	40
Saltine-type crackers	6
Starchy Foods Prepared with Fat	
Pancake (4” across)	1
Waffle (4 1/2” across)	1
Taco Shell (6” across)	1
French Toast	1

Dairy Products (90 calories per serving) Children under 2 years of age should be given whole milk.	**Serving Size**
Skim milk Soy milk (unsweetened) Rice milk (unsweetened) Almond milk Hemp milk (unsweetened) Non-fat yogurt (check label for calories)	8 oz 8 oz 8 oz 8 oz 8 oz 4-6 oz

Fat (45 calories, 5 grams of fat per serving)	**Serving Size**
Margarine/oil/mayonnaise Diet Margarine/diet mayonnaise Salad Dressing Reduced Calorie Salad Dressing Peanuts Seeds, Pine Nuts, Sunflower (without shells) Bacon Cream Cheese	1 teaspoon 2 teaspoons 1 tablespoon 2 tablespoons 20 small or 10 large 1 tablespoon 1 slice 1 tablespoon

Fruit (60 calories per serving)	**Serving Size**
Fresh, Medium Fruit Berries or Melon Canned Fruit (in juice) Dried Fruit Raisins	1 1 cup ½ cup ¼ cup 2 tablespoons

Meat and Meat Substitutes	Serving Size
Lean Meat	
(55 calories, 3 grams of fat per serving)	
Cooked poultry, without skin	1 oz
Cooked lean meat	1 oz
Cooked fish	1 oz
Tuna (in water)	1 oz
Non-fat cottage cheese	¼ cup
Diet Cheese (less than 55 calories/oz)	1 oz
95% Fat-Free luncheon meats	1 oz
Medium-Fat Meats and Substitutes	
(75 calories, 5 grams of fat per serving)	
Most beef products	1 oz
Most pork products	1 oz
Skim or part-skim milk cheese:	
Ricotta	¼ cup
Mozzarella	1 oz
Diet Cheese (56-80 calories/oz)	1 oz
Egg	1
Tofu	4 oz
High-Fat Meat and Substitutes	
(110 calories, 8 grams of fat per serving)	
Pork spareribs and sausage	1 oz
All fried fish products	1 oz
All regular cheese	1 oz
Luncheon meat- bologna, salami	1 oz
Turkey or Chicken hotdog	1
Peanut Butter	1 tablespoon

Notes

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PRADER-WILLI SYNDROME
MEDICAL ALERTS

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

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REVISED 2022

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

PWSA | USA Mission

To enhance the quality of life of and empower those affected by Prader-Willi syndrome.

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Find more valuable information on PWSA | USA's website, including a downloadable version of this Medical Alerts Booklet, by scanning the QR code below with your mobile device.

Prader-Willi Syndrome Medical Alerts
by
Clinicians of the PWSA I USA Clinical Advisory Board
and consultant experts

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INTRODUCTION

Characteristics and Common Medical Complications of PWS

This booklet was developed to alert medical practitioners in emergency departments, urgent care facilities and primary care practices to **severe medical complications that can develop rapidly in individuals with Prader-Willi syndrome (PWS).**

The booklet highlights medical issues that occur in some patients with PWS and hopefully assists in the recognition and management of problems that are uncommon in the general population but do occur with increased frequency among individuals with PWS. These findings may present at various ages and result in serious, sometimes urgent or even fatal outcomes. Common problems during hospitalization and medical procedures are also discussed.

The booklet also serves to alert families and other caregivers to potential PWS complications requiring specific management.

PWS is a variable and complex genetic neurobehavioral disorder resulting from an abnormality on chromosome 15. PWS occurs in approximately 1:10,000 to 1:15,000 births.

PWS affects the functioning of the hypothalamus and other aspects of the brain, and typically causes the following frequent findings:

- Generalized hypotonia evident prenatally and throughout life
- Decreased ability to suck in infancy leading to failure to thrive if not compensated. Swallowing abnormalities of oral secretions and food in all ages, often unrecognized
- Hyperphagia due to hypothalamically-driven lack of sense of satiety that can lead to dramatically excessive eating and, coupled with body composition abnormalities and metabolism with low caloric needs, can result in morbid obesity. Hyperphagia begins as early as ages 2-4 years and lasts throughout life. The presence of obesity can result in typical complications not usually present in those who are not obese, such as diabetes mellitus. Those with PWS who are not obese have had food intake carefully controlled by others.
- Short stature for the family if not treated with growth hormone
- Hip dysplasia, scoliosis, osteoporosis
- Delayed and incomplete sexual development
- Developmental delay and usually mild to moderate learning/cognitive deficits
- Chronic and significant problem behaviors; frank mental health conditions in some

In addition, some of the other common findings that may cause difficulties include:

- Adverse reactions to medications including anesthetics
- High pain tolerance leading to unsuspected issues such as fractures
- Gastrointestinal issues including decreased ability to vomit and chronic constipation. Occasional stomach necrosis and rupture often following binge eating
- Respiratory abnormalities such as hypoventilation or sleep disordered breathing in the form of obstructive or central sleep apnea
- Sleep problems such as excessive daytime sleepiness

- Temperature regulation abnormalities (hypothermia or hyperthermia)
- Misunderstanding or misinterpretation of information, necessitating clear and simple instructions

These findings are explained in more detail in the following pages, along with recommendations for evaluation and treatment for some of the problems in PWS. Information is based on literature review and experience of experts on PWS. The most emergent issues are discussed in the first section, inpatient, surgical and acute medical concerns in the second section, and additional medical issues and elaborations of some issues in the third section. A brief description of the genetic basis of PWS and how PWSA | USA can help in the event of death follow in sections IV and V.

Genetic testing is available for confirmation of diagnosis and to distinguish the three common causative genetic changes, which have a few distinctive findings (please see page 35 of this booklet, the section on Genetics.)

Recommended additional resources on medical issues in PWS include UpToDate® (www.uptodate.com) and GeneReviews (https://www.ncbi.nlm.nih.gov/books/NBK1330/). Members of the PWSA | USA Clinical Advisory Board are available for consultation with physicians through the Prader-Willi Syndrome Association | USA.

I. Emergency and Acute Medical Issues

Obesity and its related complications are the major causes of morbidity and mortality in Prader-Willi syndrome (PWS). Keeping the individual at a healthy weight will minimize these complications, but there are important medical and behavioral/mental health problems unique to PWS regardless of weight status.

Note that people with PWS have cognitive disability, and though it is usually mild they may misinterpret what is asked of or told to them. Instructions should be kept clear and simple.

Medical professionals can contact PWSA | USA to obtain more information and be put in touch with a specialist, as needed. UpToDate® (www.uptodate.com) and GeneReviews (https://ncbi.nlm.nih.gov/books/NBK1330) have excellent summaries of the syndrome.

Severe Gastrointestinal Concerns

- **Vomiting – Decreased ability to vomit.** Vomiting occurs infrequently in many people 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 new onset vomiting or vomiting accompanied by loss of appetite or lethargy may**

signal a life-threatening illness and may warrant immediate treatment. (See below and pages 24-25 as well as foldout on the last page of this booklet for more information on this topic.)

- **Severe Gastric Illness:** Gastric problems are very common in PWS due to decreased motility and gastroparesis. Abdominal distension or bloating, pain and/or vomiting may be signs of life-threatening gastric dilation, inflammation or necrosis. Rather than localized pain, there may be a general or vague feeling of being unwell. Anti-diarrheal medications may also cause severe colonic distension, necrosis and rupture and should be avoided. Any individual with PWS with these symptoms needs immediate medical attention. An X-ray, CT scan or ultrasound can help with the diagnosis and confirm if there is gastric necrosis and/or perforation.

  If distension is noted, these individuals need close clinical monitoring on an ongoing basis, to be made NPO, and may need decompression with an NG tube. **Gastric necrosis or perforation is a medical emergency requiring exploratory laparotomy or emergent surgery. Individuals with PWS may not have tenderness, rigidity or rebound normally associated with an acute abdomen.** Please see additional information on Gastric Necrosis on pages 24-25 of this booklet and see *an algorithm for evaluation of GI complaints in people with PWS at the end of this booklet.* See also **http://www.pwsausa.org/resources/medical-issues-a-z/** and view GI Problems-stomach and intestines.

- **Constipation and Rectal Bleeding:** Although only 20% of adults with PWS report constipation, a recent study found that 40% of adults with PWS fulfilled the diagnostic criteria for constipation. Abdominal and rectal pain, rectal fissures, and rectal bleeding may occur in association with disordered defecation. Rectal ulcers have occurred in individuals with PWS as a result of localized deep rectal picking aggravated by rectal irritation from constipation/anal pruritus and can present with mucoid rectal discharge, bloody stools, rectal pain, and tenesmus suggestive of emerging inflammatory bowel disease and warranting gastroenterology consultation. **Colonic impaction** may also occur and needs to be addressed. See also Constipation on pages 25-26 and at **http://www.pwsausa.org/resources/medical-issues-a-z/**

- **Other GI issues**: Stomach pain can also be due to gallstones or pancreatitis. An ultrasound, chemistry analysis of the blood and CT of the abdomen will help with the diagnosis.

Swallowing Dysfunction and Choking

People with PWS are highly likely to have an undetected swallowing problem that places them at risk for asphyxiation by a food bolus (choking). Many people with PWS cannot tell if they have cleared their throat or airway after swallowing, increasing the risk for aspiration. As a result, assessment requires a special kind of evaluation, a videofluoroscopic swallowing study with an esophageal

sweep. A clinical or bedside evaluation is not sufficient to detect dysphagia in this population. Choking can also occur with rapid ingestion of food and has caused numerous deaths in the PWS population. For more information on this topic, please see **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Choking/Swallowing.

Respiratory Concerns

Individuals with PWS are at increased risk for respiratory difficulties. They have blunted ventilatory responses to hypoxemia and hypercarbia. This can cause problems related to anesthesia and sedation and complicate the diagnosis of obesity hypoventilation syndrome. Hypotonia, weak chest muscles, swallowing abnormalities, and central or obstructive sleep apnea are common. Anyone with significant snoring or other sleep problems, regardless of age or presence of obesity, should have a medical evaluation to look for sleep disordered breathing. This may include a sleep study. Infants commonly have central sleep apnea which generally improves spontaneously over time, but they may also have obstructive sleep apnea due to the hypotonia and other factors, as may individuals with PWS of all ages. Hypotonia can lead to diminished activity levels and low aerobic capacity. People with PWS at all ages are at risk for hypoventilation, which is central in origin. Hypersomnolence with or without cataplexy has been described in PWS.

In children with PWS, chronic stomach reflux and aspiration are emerging as common problems. Reflux should be considered in young children with chronic respiratory problems; videofluroscopy is the preferred test. Individuals with obstructive sleep apnea or obesity are at higher risk for reflux. At any age morbid obesity can be associated with obesity-hypoventilation syndrome. Children with PWS have been shown to have hypoventilation disproportionate to obstructive sleep apnea. (Please see pages 26-30 for recommendations for evaluation of breathing abnormalities associated with sleep disorders.)

Medications – Adverse Reactions

People with PWS may have unusual reactions to standard dosages of medications. Use extreme caution in giving medications, especially those that may cause sedation; prolonged and exaggerated responses have been reported. Metabolism of the drugs may be impaired in individuals with PWS. Abnormal body composition and metabolism may affect pharmacokinetics. In obese individuals, weight-based dosing guidelines often do not specify the use of actual body weight versus ideal or adjusted weight estimates, and multiple additional factors impacted by obesity must be considered for appropriate dosing. Consider additional focus on renal and hepatic function, medication lipophilicity, recommended dosing weight, and observability of medication effects. Special care should be taken with medications that have a narrow therapeutic window and for those in which the detection of harm may be delayed.

Pain Tolerance

A high threshold for sensing pain 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. Any complaint of pain by a person with PWS should be taken seriously.

Skin Picking and Bruises

Because of a compulsion that is common in PWS, open sores caused by skin picking may be apparent. Rectal picking/gouging is not uncommon. Individuals with PWS also tend to bruise easily. These lesions can cause serious life-threatening infections. Appearance of such wounds and bruises may erroneously lead to suspicion of physical abuse. There are approaches to help mitigate picking. Please see **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Skin Picking.

Falls and Fractures

Individuals with PWS may have significant fractures from simple falls and require X-rays even if they do not complain of pain. Persistent pain, swelling, guarding, limping, or decreased movement of an extremity for more than a few days may warrant an X-ray.

Water Intoxication

Water intoxication has occurred in relation to use of certain medications with antidiuretic effects, as well as from excess (binging) fluid intake alone. For additional information see **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Water Intoxication.

Temperature Abnormalities

Idiopathic hyperthermia and hypothermia can be noted in people with PWS. Hyperthermia may occur during minor illness and in procedures requiring anesthesia. Fever of unknown origin occurs. However, malignant hyperthermia does not appear to occur at increased frequency in PWS. On the other hand, fever may be absent despite serious infection. All individuals with PWS are at risk for mild hypothermia because of impaired peripheral somatosensory and central thermoregulation, poor judgment and cognitive inflexibility. Hypothermia is common in infants with PWS. See **http://www.pwsausa.org/resources/medical-issues a-z/** and view Temperature and Hypothermia.

Central Adrenal Insufficiency

Central adrenal insufficiency is a rare occurrence in people with PWS. A stress dose of cortisol may be indicated if the individual has problems after surgery or during times of stress. See page 34 in this booklet for more information. See also **http://www.pwsausa.org/medical-issues-a-z/** and view Adrenal or Cortisol Insufficiency.

Hyperphagia and Food Seeking

Individuals with PWS have a nearly constant drive to eat and must be continuously supervised in all settings to prevent access to excess food. In hospital settings, obtaining unguarded food can lead to rapid ingestion and fatal choking or gastrointestinal issues. Individuals who have normal weight have achieved this because of strict external control of their diet and food intake; these individuals are not less likely to ingest available food. There are currently no treatments for this constant urge to eat. Insatiable appetite may lead to life-threatening weight gain, which can be very rapid and occur even on a low-calorie diet.

II. Peri-Operative and In-Patient Issues

A. Hospital Experience and Pre-Anesthesia

Pre-Operative Preparation

When possible, pre-operative preparation to optimize nutritional status and address the common problems of diabetes control issues and constipation prior to significant elective surgical procedures should occur in patients with Prader-Willi syndrome.

Obesity Complications

A common finding in people with PWS, obesity can cause obstructive sleep apnea, pulmonary hypertension, diabetes, and right heart failure. These should be sought and addressed, as they affect illness, surgical and post-operative management.

Venous Access Difficulties

Many people with PWS will have difficult intravenous (IV) access due to increased fat mass and smaller than normal blood vessels. Ultrasound guided peripheral IV placement is helpful. IV lines are often more distressing to children with PWS than their actual surgery, therefore the lines need to be protected. In situations where hydration for more than 2-3 days is required, consider a peripherally inserted central catheter (PICC line) or tunneled central venous access, to avoid reinsertion.

Hyperphagia/Food Seeking

For people with PWS, complete safety from access to extra food is essential in any health care setting. Access to food storage or refrigerators should be prevented. Assume the individual has eaten unless verified by a caregiver. Complaints of hunger should not result in access to snacks or food. Patients in the hospital should have someone with them at all times. The individual may be on a calorie-restricted diet, and that should be conveyed to the nutritionist and kitchen. For elective procedures involvement of a dietician to help with pre-operative nutritional management along with planning for inpatient management of nutrition may be helpful. Patients with PWS should not be permitted to have "at your request" or "on demand" food ordering. A dietician should be involved in setting up the inpatient and discharge nutrition plans to ensure adequate protein intake along with appropriate vitamin/mineral supplementation to provide optimal healing.

Pain Tolerance

Unexplained tachypnea or tachycardia may be the only indication of pain. Behavior problems that are not typical for this person may also be evidence of pain. Individuals with PWS may not respond to pain in the same manner as others, masking the presence of underlying problems. Since pain may not be evident, other signs of underlying problems should be assessed.

Behavior Problems

Individuals with PWS are prone to emotional outbursts, obsessive-compulsive behaviors, and psychosis. These may be exacerbated by the stress of hospitalization or surgery. If possible, a pre-admission assessment should be performed, in part to consider 1-to-1 supervision in order to safeguard staff and the patient and prevent food foraging.

Psychosis

There is an increased risk of psychosis in individuals with PWS, which can be triggered by significant events such as changes in routines or serious illness. Prompt attention to hallucinations or reported change in typical behavior is essential. View Psychiatric Concerns at **http://www.pwsausa.org/resources/medical-issues-a-z/**

Skin Picking

Picking at sores and stitches is a common self-injurious behavior in PWS. It may complicate healing of IV sites and incisional wounds. Restraints or gloves may be necessary to protect wounds during healing. See Skin Picking at **http://www.pwsausa.org/resources/medical-issues-a-z/**

Temperature Instability

Low basal body temperature is typical in healthy individuals with PWS. Hypothalamic dysregulation can lead to poor temperature control during fever or hypothermia.

Respiratory Issues

The high incidence of central, obstructive and mixed apnea in people with PWS make it imperative to obtain a sleep study and/or pulmonology consultation prior to moderate or major surgical procedures in order to guide post-operative use of CPAP or BiPAP. The generalized hypotonia may include respiratory muscle weakness, which could complicate the ability to cough effectively and clear airways. See pages 26-30 in this booklet and **http:pwsausa.org/resources/medical-issues-a-z/** and view Breathing/ Respiratory concerns.

Cardiac Problems

Surprisingly, coronary disease is less in PWS than in individuals with similar obesity. Cardiac problems, if they do occur, usually are due to hypoventilation right heart failure, which can be associated with obesity. Non-pitting edema can often be seen in the obese individual even in the absence of heart failure and is treated with weight loss and ambulation. Diuretics are usually not very beneficial in treating the edema.

B. Anesthesia and Surgical Procedures

General Recommendations

Schedule procedures as early in the day as possible to prevent prolonged awake NPO status, so as to reduce patient anxiety and opportunities for food seeking behavior.

Anesthesia

People with PWS may have unusual reactions to standard dosages of anesthetic agents. Use caution in giving anesthesia. Outpatient procedures and conscious sedation may be especially problematic; the use of general anesthesia and airway management is often preferred but may warrant overnight observation for respiratory complications. Procedures done outside of the hospital settings should be carefully considered, with proper equipment for resuscitation immediately available. Ongoing assessment of breathing and oxygen saturation is critical in all outpatient procedures including dental work. Ongoing psychotropic medications may affect metabolism of anesthetic agents leading to shorter or longer duration of action. People with PWS may exhibit abnormal physiological responses to hypercapnia and hypoxia. There does not seem to be a higher incidence of malignant hyperthermia. Please see **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Anesthesia**.**

Narcotic Sensitivity

Individuals with PWS may have an exaggerated response to narcotics. Use the lowest possible dose to achieve the desired state of anesthesia. Many individuals with PWS have delayed gastric emptying that can be compounded with narcotics.

Airway Access

A small airway, high palate, and/or obesity (neck and pharyngeal adiposity) may complicate ability to intubate. It can also make bag-mask ventilation difficult (mask fit challenges, increased airway resistance and reduced respiratory system compliance). Outpatient procedures and general sedation may be especially problematic. Care must be taken during procedures done in or out of hospital settings, and assurance that proper equipment for resuscitation is immediately available if needed. The possibility of doing such procedures in an operating room should be discussed. Procedures where more than light sedation is used may warrant overnight observation, particularly since sensitivity to medications is also an issue in PWS (see below).

Saliva Abnormalities

Thick sticky saliva complicates airway management especially during conscious sedation. It also increases the risk of caries. Dried saliva may not be an indication of hydration status. Voluntary water drinking is minimal in the majority of individuals with PWS.

Oro-Pharyngeal Surgical Concerns

With a significant number of infants and children with PWS undergoing sleep assessments prior to growth hormone treatment and the potential consequent rise in surgical procedures (e.g., tonsillectomy) requiring intubation and anesthesia, it is important to alert the medical team about complications. These may include trauma to the airway, oropharynx or lungs due to possible anatomic and physiologic differences seen in PWS, including a narrow airway, underdevelopment of the larynx and trachea, hypotonia, edema, and scoliosis.

C. Post-Operative Period

General Recommendations

Patients with PWS who undergo deep sedation or general anesthesia should be recovered overnight in a monitored unit. Continuous monitoring of pulse-oximetry for 24 hours is important post-operatively, with attention to airway and breathing. Infants and children may require intensive care monitoring. A conservative approach to pain management should be used, limiting the use of narcotic agents. Consider direct supervision (1:1) for those patients at risk of food foraging post-operatively. Patients may exhibit altered temperature regulation, where fevers may be absent despite the presence of infection. Individuals with PWS are at risk for deep venous thrombi (DVT) and pulmonary embolism due to their hypotonia and obesity. DVT prophylaxis should be considered in all obese individuals with PWS, and prolonged bed rest is to be avoided. Please review the sections above under Hospital experience and Pre-Anesthesia (pages 12-15) and **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Post-operative Monitoring.

Respiratory Considerations

Pre-operative pulmonary assessment should guide the use of CPAP or BiPAP. Respiratory therapy may be indicated to prevent atelectasis and/or post-operative lung infections.

Pain Insensitivity and Narcotics

Individuals with PWS characteristically display a decreased outward response to pain. The only indications of pain may be behaviors that are not typical for that individual, or unexplained tachypnea/tachycardia. Lack of a typical pain response may mask the presence of underlying problems. Conversely, many post-surgical patients with PWS seem to experience less pain, and they can be comfortable with lower doses of narcotic medications or with a narcotic-free regime. Those who do need post-operative narcotics may benefit from methylnaltrexone to decrease the duration of the post-operative ileus.

Gastrointestinal Issues

Post-operative ileus is characteristically more profound and long lasting in patients with PWS. When indicated, sips of clear liquids may be started immediately after surgery, but the advancement of diet should be delayed until there are non-subjective signs of digestive recovery. One strategy for moderate to extensive surgeries on older children or adults is 2 ounces of clear liquids every 4 hours to start. If the patient tolerates intake and bowel sounds are present, the intake can be increased to 4 ounces every 4 hours. Abdominal radiographs are done daily to confirm normal gas patterns before advancing to a soft diet. Any abdominal bloating is an indication to discontinue diet.

Skin Picking

Skin picking may represent a severe threat to post-operative incisions. Restraints or gloves may initially be necessary, followed by physical barriers such as braces or casts to protect wounds during healing. Post-operative anxiety may cause patients without a history of skin picking to begin the habit.

Hypotonia Consequences

Generalized muscle hypotonia is a constant feature of PWS. It may complicate ability to cough effectively and clear airways, affecting post-operative recovery.

Pulmonary Embolism

Individuals with PWS are at increased risk for pulmonary embolism. Deep vein thrombosis prophylaxis should be considered in all obese individuals. Prolonged bed rest should be avoided.

Orthopedic Concerns

Musculoskeletal manifestations, including scoliosis, hip dysplasia, fractured bones (which may be undetected), osteoporosis, and lower limb alignment abnormalities, occur at significant frequency in people with PWS. Care of this patient population from the orthopedic surgeon's

perspective is complicated by other clinical manifestations of PWS. Please see also **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Orthopedic Issues**.**

Behavioral Disorder and Psychosis

People with PWS are prone to emotional outbursts, obsessive-compulsive behaviors, and in some cases psychosis. Psychosis can be triggered by significant events such as changes in routines and serious illness. Prompt attention to hallucinations, disorientation or reported change in typical behavior is essential. View mental health issues at **http://www.pwsausa.org/resources/medical-issues-a-z/** under Psychiatric Concerns.

Summary of Post-Operative Management

Patients with PWS are known to have increased morbidity after surgery due to:

- Abnormal physiological response to hypercapnia and hypoxemia
- Untreated central and/or obstructive sleep apnea
- Hypotonia
- Narrow oropharyngeal space
- High incidence of central, obstructive and mixed apnea
- Thick secretions
- Obesity
- Increased incidence of scoliosis with decreased pulmonary function

- Prolonged exaggerated response to sedatives
- Increased risk for aspiration
- Decreased pain sensation
- Possible challenges with compliance to pre- and post-operative treatment procedures due to:
  - Extreme food seeking behavior and hyperphagia
  - High incidence of gastroparesis and slow motility of the intestinal tract
  - Severe skin picking which may interfere with wound healing
  - Altered temperature regulation – fever may be absent in the presence of infection
  - The possibility of central adrenal insufficiency

Therefore, the following are recommended for post-operative management:

- Patients with PWS who undergo deep sedation and general anesthesia should be recovered overnight in a monitored unit. Infants and children may require intensive care monitoring.
- Continuous monitoring of pulse-oximetry for 24 hours post-operative with attention to airway and breathing.
- A conservative approach to pain management and use of narcotic agents.
- Full assessment of return of gastrointestinal motility prior to initiation of intake by mouth, often with abdominal radiographs, because of the predisposition to ileus after surgery.

- Scheduling procedure as early in the day as possible to prevent prolonged time period where food seeking could take place.
- Direct supervision (1:1) to prevent foraging post-operatively and exclusion from ad lib patient ordering of food from hospital dietary services.
- Monitor for picking at wounds and/or incisions. These may require additional dressings and other barriers including full time sitter to prevent access to surgical site and medical devices
- Close observation of wound for signs of infection
- Airway clearance to prevent atelectasis and/or post-operative lung infection.
- Due to the hypotonia and obesity, individuals with PWS are at risk for deep venous thrombi (DVT) and pulmonary embolism. Patients should be under the guidelines for DVT prophylaxis.

Please see also **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Post-operative/Surgery.

III. Evaluation and Treatment of Special Issues

Risk of Stomach Necrosis and Rupture

A Cause of Death from Sepsis, Gastric Necrosis or Blood Loss

Signs and symptoms of stomach necrosis and rupture:

- **Vomiting-**Atypical vomiting accompanied by decrease in appetite or lethargy is unusual in PWS
- **Loss of appetite** (ominous sign)
- **Lethargy**
- **Complaints of pain, usually non-specific.** Pain sensation appears to be abnormal in PWS due to high pain threshold; affected people rarely complain of pain
- **Pain** is often poorly localized
- **Peritoneal signs** may be absent
- **Abdominal/stomach bloating** and **gastric dilation**
- **Fever** may or may not be present
- **Guaiac positive stools (chronic gastritis)**

An algorithm for Emergency Room evaluation of an individual with PWS and abdominal complaints is on a foldout page at the back of this publication.

History may include:

- History of **binge eating** within the week. Hyperphagia and binge eating are characteristic of people with PWS, regardless of whether obese or slim. This frequently occurs at holiday or social occasion with less supervision of intake
- History of **gastroparesis**, which is common in PWS, though often undiagnosed
- History of significant **obesity followed by weight loss**, which may leave the stomach wall thinned.
- See also **http://www.pwsausa.org/resources/medical-issues-a-z/** under GI Problems**.**

Constipation

Constipation is a common problem in individuals with Prader-Willi syndrome (PWS). Although only 20% of adults with PWS report constipation, a recent study found that 40% of adults with PWS fulfilled the diagnostic criteria for constipation. It takes longer for food to move through the GI system (gastroparesis) in Prader-Willi syndrome. This slower passage of food can lead to serious issues similar to the ones seen related to the stomach. Outpatient methods used to clear constipation in non-PWS patients may be ineffective due to poor fluid intake and hypotonia. Inpatient regimens frequently use large volumes of fluid which may cause problems. Reliance on these methods may lead to life-threatening conditions such as necrosis and perforation of the colon and subsequent sepsis. Due to decreased muscle tone and altered pain response, individuals with

PWS may not have the same clinical exam that a non-PWS patient would have. A heavier reliance on imaging may be necessary. Individuals with PWS may be at higher risk for impaction. Rectal examination and enema may be required in addition to oral cleanout regimen. This may also be problematic in some, leading to rectal picking.

Patients with PWS having constipation and receiving repeated regimens of oral PEG (polyethylene glycol) solution for bowel cleansing should be monitored closely for abdominal distention and retention. Use of laxative agents with sweeter flavoring, such as lactulose or chocolate-flavored senna preparations, should be avoided if possible.

Failure of standard constipation protocols to clear the stool in a timely manner, especially in the face of increasing abdominal distention, vomiting, decreased appetite, stoppage of food consumption, and/or abdominal pain, warrants surgical or GI consultation. Emergent surgical or colonoscopic intervention may be necessary.

Breathing Abnormalities Associated with Sleep

Problems with sleep and sleep disordered breathing have long been known to affect individuals with PWS. The problems have been frequently diagnosed as sleep apnea (obstructive [OSA], central or mixed) and/or sleep related hypoventilation with hypoxemia. Disturbances in sleep architecture (delayed sleep onset, frequent arousals and increased time of wakefulness) are also frequent. Sleep

problems in people with PWS are often underrecognized as they do not exhibit the most common symptoms such as snoring, witnessed apneas, etc.

Factors that seem to increase the risk of sleep disordered breathing include young age, severe hypotonia, narrow airway, morbid obesity, and prior respiratory problems requiring intervention such as respiratory failure, reactive airway disease and hypoventilation with hypoxemia. Due to a few fatalities reported in individuals with PWS who were on growth hormone therapy (GH), some physicians have also added this as an additional risk factor. One possibility (that is currently unproven) is that GH could increase the growth of lymphoid tissue in the airway thus worsening already existing sleep disordered breathing. Nonetheless, it must be emphasized that there is currently no definitive data demonstrating that GH causes or worsens sleep disordered breathing. However, to address this concern, as well as the historically well documented increased risk of sleep-related breathing abnormalities in PWS, **the Clinical Advisory Board of the PWSA | USA makes the following recommendations:**

1. **A sleep study or a polysomnogram** that includes measurement of oxygen saturation and carbon dioxide for evaluation of hypoventilation, obstructive sleep apnea and central apnea should be contemplated for all individuals with Prader-Willi syndrome. These studies should include sleep staging and be evaluated by experts with sufficient expertise for the age of the patient being studied.

2. **Risk factors that should be considered to expedite the scheduling of a sleep study should include:**
   - Severe obesity – weight over 200% of ideal body weight (IBW).
   - History of chronic respiratory infections or reactive airway disease (asthma).
   - History of snoring, sleep apnea or frequent awakenings from sleep.
   - History of excessive daytime sleepiness, especially if this is getting worse.
   - Before major surgery including tonsillectomy and adenoidectomy.
   - Prior to sedation for procedures, imaging scans and dental work.
   - Prior to starting growth hormone or if currently receiving growth hormone therapy.

Additional sleep studies should be considered if patients have the onset of one of these risk factors, especially a sudden increase in weight or change in exercise tolerance. **If a patient is being treated with growth hormone, it is not necessary to stop the growth hormone before obtaining a sleep study unless there has been a new onset of significant respiratory problems.**

Any abnormalities in sleep studies should be discussed with the ordering physician and a sleep specialist knowledgeable about treating sleep disturbances to ensure

that a detailed plan for treatment and management is made. Referral to a pediatric or adult sleep medicine specialist is strongly encouraged for management of the respiratory care.

In addition to a calorically restricted diet to ensure weight loss or maintenance of an appropriate weight, a management plan may include modalities such as:

- Supplemental oxygen
- Continuous positive airway pressure (CPAP) or Bilevel positive airway pressure (BiPAP)
- Oxygen should be used with care as some individuals may have hypoxemia as their only ventilatory drive and oxygen therapy may actually worsen their breathing at night.
- Behavior modification therapy is sometimes needed to gain acceptance of CPAP or BiPAP.
- Medications to treat behavior may be required to ensure adherence to the treatment plan.

If sleep studies are abnormal in the morbidly obese child or adult (>200% IBW), the primary problem of weight should be addressed with an intensive intervention — specifically, an increase in exercise and dietary restriction. Both are far preferable to surgical interventions of all kinds. Techniques for achieving this are available from clinics and centers that provide care for PWS and from the national parent support organization [PWSA | USA]. Behavioral problems interfering with diet and exercise may need to be addressed simultaneously by people experienced with PWS.

If airway related surgery is considered, the treating surgeon and anesthesiologist should be knowledgeable about the unique pre- and post-operative problems found in individuals affected by Prader-Willi syndrome.

Tracheostomy surgery and management present unique problems for people with PWS and should be avoided in all but the most extreme cases. Tracheostomy is typically not warranted in the compromised, morbidly obese individual because the fundamental defect is virtually always hypoventilation, not obstruction. Self-endangerment and injury to the site are common in individuals with PWS who have tracheostomies placed.

At this time there is no direct evidence of a causative link between growth hormone and the respiratory problems seen in PWS. Growth hormone has been shown to have many beneficial effects in most individuals with PWS including improvement in the respiratory system. Decisions in the management of abnormal sleep studies should include a risk/benefit ratio of growth hormone therapy. It may be reassuring for the family and the treating physician to obtain a sleep study prior to the initiation of growth hormone therapy and after 6-8 weeks of therapy to assess the difference that growth hormone therapy may make. A follow-up study after one year of treatment with growth hormone may also be indicated.

Endocrine Abnormalities

Hypothalamic dysfunction and its resultant hormone deficiencies are the presumed origin of many features of PWS.

- **Hypothyroidism** (thyroid stimulating hormone deficiency) has been reported to occur in up to 20%-30% of individuals and may be undiagnosed prior to surgery. Central and primary hypothyroidism can be seen in individuals with PWS. Levothyroxine treatment should not be routinely prescribed in children with PWS unless confirmed by thyroid function testing. Both plasma thyroid stimulating hormone (TSH), T_4 and free T_4 (FT_4) are low in central hypothyroidism, whereas TSH is elevated in primary hypothyroidism. It is recommended that baseline thyroid function testing (T_4/FT_4 and TSH) be done during the first 3 months of life (unless the newborn screening was normal) and annually thereafter, especially if the patient is receiving GH therapy. Please see Hypothyroidism at **http://pwsausa.org/resources/medical-issues-a-z/**

- **Growth hormone deficiency** is also related to hypothalamic dysfunction. All individuals with PWS should be considered to be growth hormone (GH) deficient. Currently, growth hormone is being used as early as one month of life with overall beneficial effect on body composition and growth. The recommended dose is 0.18 to 0.24 mg/kg of ideal body weight divided 7 days a week. The lowest dose is recommended in infants. Benzyl alcohol free-GH products such as Genotropin Miniquick should be first choice whenever possible during the first 6 months of life. Bone age, growth velocity, plasma IGF-1, $IGFBP_3$, glucose, HbA1C, insulin, and thyroid function testing should be monitored during GH treatment.

  Overall, GH therapy is generally safe and well tolerated in PWS children and adolescents. Extreme caution, however, is recommended during 3-12 weeks after initiating GH due to possible development of increased intracranial pressure, manifested by headache and papilledema. It resolves by stopping GH and restarting thereafter with low GH dose with gradual increase. Due to possible development of obstructive sleep apnea, polysomnography should be obtained prior to initiating treatment, within 3-6 months after starting GH therapy, and then annually. Scoliosis is not a contraindication for GH treatment.

  While GH is typically discontinued once bone maturation is achieved at a bone age of 14.5 and 16.5 in girls and boys, respectively, it is the consensus of experts that GH remains beneficial throughout the lifespan. An adult GH stimulation test is necessary to consider adult GH treatment. GH dose in adults is 0.2 to 1.2 mg daily. Lower extremity edema is the most common side effect, but it subsides after decreasing the GH dose. The same blood work as for children is needed to monitor GH treatment in adults, with bone mineral density instead of bone age. See Growth Hormone at **http://pwsausa.org/resources/medical-issues-a-z/**

- **Hypogonadism** occurs in both sexes. Both central or hypogonadotropic (low LH/FSH) and primary or hypergonadotropic (ovarian failure) hypogonadism have been reported in PWS.

Cryptorchidism is virtually universal in males with PWS. Although human chorionic gonadotropin (hCG) is only effective in 24% of infants, this modality of treatment should be considered before a surgical approach. Early treatment with hCG may result in better outcomes including improved development of the scrotal sac, growth of phallus length and muscle tone. The improved muscle tone may decrease the need for gastrostomy tube feeding and facilitate circumcision and orchidopexy.

The increase in onset of pubic and/or axillary hair before age 8 years in girls and 9 in boys is most commonly the result of premature adrenarche and should not be confused with an early sign of puberty. Testicular enlargement (4 ml) in boys and breast development in girls is the first sign of puberty.

No consensus exists as to the most appropriate regimen for sex hormone replacement in PWS. However, most experts recommend intramuscular testosterone replacement in males starting at a dose of 25-50 mg given every 28 days, usually by age 14 years, with gradual increase towards typical adult male doses. Behavior should be monitored during treatment. Other modalities of androgen therapy include daily patches or gel as well as testosterone enanthate, which is administered subcutaneously once a week, typically administered by the parents. Oligomenorrhea or amenorrhea is typical for females with PWS. In girls, usually by age 12-13 years, low-dose oral estrogens with gradual increase are recommended, with combined oral contraceptive pills used after the first vaginal bleeding has occurred. Monitoring of sex hormone replacement therapy should include LH, FSH and sex hormones (testosterone or estrogens).

Although rare, there have been six documented pregnancies in females with PWS. Therefore, counseling on reproductive health and contraceptive practices is warranted for all females with PWS. See Puberty/Sex Hormones at **http://pwsausa.org/resources/medical-issues-a-z/**

Central Adrenal Insufficiency

The possibility has been raised of unrecognized adrenal insufficiency as the responsible cause of unexplained death in some individuals with PWS. However, subsequent studies based on various means of dynamic testing revealed low rates of central adrenal insufficiency in PWS, ranging from 0 to 14.3%. It is currently considered rare.

In general practice, the first step in evaluating patients for possible central adrenal insufficiency is measuring a morning (8 to 10 a.m.) basal cortisol level. Dynamic testing should be considered if repeat cortisol is still below normal range. None of the dynamic stimulation tests can be considered completely reliable for establishing or excluding the presence of central adrenal insufficiency. Consequently, clinical judgment remains one of the most important issues for deciding which patients need assessment or reassessment of adrenal function.

IV. Genetic Basis of PWS

PWS is due to a genetic abnormality that, in the vast majority of cases, results from a new genetic change in the person with PWS. It is caused by lack of expression of a group of genes on the proximal long arm of chromosome 15 (15q11.2-q13). In about 2/3 cases, this expression deficiency is due to absence (**deletion**) of a segment of the chromosome 15 contributed to the affected individual by the father. In most of the remaining cases it is due to the presence of two maternally-contributed and no paternally-contributed chromosome 15 (**maternal uniparental disomy**). Normally the relevant genes in the PWS 15q region are only expressed when inherited from the father and not when inherited from the mother, a process called genomic imprinting. The third, least common cause is a defect in the imprinting center such that both parental copies of the relevant genes in the PWS region of chromosome 15 are suppressed (an **imprinting defect**).

Although most of the manifestations of PWS are the same regardless of the cause of absent expression of these genes, a few problems occur more frequently in the presence of one or the other of the genetic causes leading to somewhat different prognosis. Recurrence risk can also vary with genetic cause. All three of these genetic causes will result in an abnormal DNA methylation test, though determination of the exact genetic cause requires additional testing. Further discussion of the genetics of PWS and the manner in which they can influence medical problems is beyond the scope of this brochure but can be found in numerous sources including Gene Reviews: **https://ncbi.nlm.nih.gov/books/NBK1330/**

V. In the Event of Death

PWSA | USA collects information on mortality to help advance knowledge about causes of death. That knowledge can lead to future research and address critical areas of need for advancing treatment development and quality of life/longevity for those with PWS. We also partner with Autism BrainNet to facilitate the collection of postmortem brain tissue to identify new and effective treatments.

When the death of a loved one is near or has occurred, families may call Autism BrainNet's 24-hour, seven-day-a-week hotline to begin the donation process:

877.333.0999 or PWSA | USA **941.312.0400**

PWSA | USA also provides bereavement support to families who have lost a child with PWS. Please call PWSA | USA to report a death so that the family can receive grief counseling. Please contact Family Support (**941.312.0400**) in the event of death/near-death of an individual with PWS.

Prader-Willi Syndrome Association | USA
941.312.0400 | info@pwsausa.org | wwww.pwsausa.org

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Prader-Willi Syndrome Medical Alerts
by
Clinicians of the PWSA | USA Clinical Advisory Board
and consultant experts

This life-saving Medical Alerts Booklet is dedicated to ***Janalee Heinemann, MSW*** *in appreciation for a lifetime of service to the PWS community and the truly thousands of lives that were saved and transformed by her skill, compassion, and dedication.*

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