Prader-Willi
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FOOD IN THE CLASSROOM

Prader-Willi Syndrome (PWS) is a genetic disorder that affects appetite, growth, metabolism, cognitive functioning, and behavior. Chronic feelings of insatiable hunger and slowed metabolism are present. Therefore,people with PWS metabolize their food at half the rate as their peers do and can easily gain weight when consuming food outside of their daily scheduled meals.

Many school districts have courses embedded in their curriculum that involve food. Additionally, teachers mayincorporate instructional activities and celebrations with food into their daily lessons. For students with Prader- Willi Syndrome (PWS), including food into the classroom may impede their learning and cause behavioral issues inall environments. Here are some helpful tips for the classroom.

- Provide food security. Food security means the ready availability of nutritionally adequate and safe foodswith an assured ability to acquire these foods in socially acceptable ways. This means students with PWSwill only access food during scheduled meals and snack times.
- Lock all access to food. This may include locking teacher's desks or cabinets in the classroom that containfood items. It may also include putting other student's lunches into a locked container that is taken to the cafeteria for other students to access during their scheduled lunch time.
- Provide a structured predictable routine for all mealtimes.
- Person with PWS should not be "punished" for obtaining unlocked food.
- Food should not be used as a reward.
- Provide close supervision or 1:1 adult support during the school day.
- Provide an alternative setting for scheduled mealtimes if necessary.

All students with Prader-Willi Syndrome are different. There are some students that may be able to handle foodexposure if extensive planning and preparation occurs in advance with the school team and the parents.

- Notify the parents in advance of food activities that will occur. For example, if a classroom pizza party isplanned, the parent could send in an appropriate food replacement or a piece of pizza that follows the student's specific diet plan. Parents may also decide to make trade-offs so that the student can enjoy a special treat at school by adjusting the lunch or dinner menu.
- Prepare the student for food exposure that is not typical (field trips, special events, parties) by using visualsand schedules. The visuals should show exactly what food will be provided and at what time.

At the middle school and high school level, many students participate in classes focused on learning a range of daily living skills, including personal care, money skills, community use, and home living skills which encompassmeal planning and preparation. While all of these areas are of value, community utilization, when it involves access to food, should be planned with attention to what the student with PWS can handle.

- If it's a field trip that includes a snack or lunch at a restaurant, the teacher should coordinate with the parent(s) regarding the details so a specific plan can be developed regarding exactly what food would bepermitted and the degree of supervision required.
- If the class is focused on food planning and preparation, the teacher and parent(s) need to discuss what thatwould consist of and whether or not the student would be appropriate for such a class.

- If the food being prepared can be incorporated in the student's meal plan, perhaps there could be somedegree of participation.
- Oftentimes an alternative activity is warranted; one which is meaningful for the student, so s/he doesn't feellike s/he is totally missing out!
- Students with PWS are not good candidates for vocational training experiences where food is involved oreasily accessible. This includes restaurant work of any kind, grocery stores, food pantries, etc.
- Vocational activities that have been successful for persons with PWS include flower shops, pet stores (although pet food may be a temptation for some students), libraries or offices where clerical tasks areperformed.
- Most work places have staff break rooms which must be carefully supervised when the student is in closeproximity or is utilizing them.

Questions for School Teams

These questions are helpful to ask as a team when preparing to support a person with PWS in theschool environment.

- Is there a breakfast program at the school where students eat in the classroom?
- Are there scheduled snack times in the classroom?
- Where are student lunches and snacks kept?
- Are students allowed to keep food, water or other liquids on their desk throughout the day?
- Do the teachers and support staff drink coffee, water, other liquids throughout the day?
- If food is kept in the classroom, is it locked in a cabinet?
- If the teacher keeps a food stash, is it locked in a drawer or cabinet?
- Does the school provide the snacks? What do they typically consist of?
- Is food used as rewards?
- Do classroom projects utilize food (macaroni pictures)?
- Are there any special classroom activities involving food: birthday parties, holiday parties(including Halloween), cultural celebrations involving food?
- Are there any field trips scheduled where food is available (zoo, shopping trips, restaurants)?
- Are cooking classes part of the curriculum?
- Does the school have an explicit policy against food sharing of any kind?

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Health Concerns and the Student w/ Prader-Willi Syndrome - Information for School Staff

The student with Prader-Willi syndrome (PWS) may experience some unique health issues. It is important for school staff to be aware of these issues to help ensure that the student has a safe, healthy educational experience. Health concerns along with some strategies are summarized below.

Health Concern	Strategies	Health Concern	Strategies
Altered Pain Threshold – Decreased Pain Sensitivity/High Pain Threshold Pain may be diminished or absent even in severe injuries. Fatigue or irritability may be a sign of illness. Increased bruising& swelling is common.	All injuries should be assessed by an adult. Report all injuries or changes in behavior to the parent or caregiver. Elevate and apply ice to injuries as needed. Student may require examination by a physician to rule out fracture or other health problem.	Skin Picking Common problematic behavior seen in students of all ages. Open sores common. May pick at various openings of body	Provide diversion activities – keep hands busy. Encourage liberal application of lotion. Incentive program often needed to keep wound covered. Teach self care of wound if able. Monitor frequent trips to bathroom. Set time limits; supervise in bathroom if needed.
Altered Temperature Regulation Common to see unexplained high and low temperatures Little or no fever may be present with illness. Often experience low tolerance to high or low outside temperatures.	Limit time outdoors during very warm and/or humid temperatures. If extreme redness of the face and sweating is noted, remove to cool area; encourage cool water and/or utilize cooling measures. In colder climates make sure student is appropriately dressed and limit exposure to cold temperatures. If illness is suspected, notify parent.	Behavior – Emotional Problems Students with PWS have problems regulating their emotions. Most do not handle change well. Some exhibit obsessive-compulsive tendencies, exaggerated emotional responses and extreme anger. Some take medications to assist with mood stabilization.	Minimize changes. When they do occur – prepare if possible. Teach ways to appropriately share feelings and emotions. Practice and reinforce these strategies frequently. State behavior you want to see. Avoid using word “don’t”. Make sure to administer medications at the appropriate times.
Increased Food Drive/Food Seeking/Low Metabolism Because of a hypothalamic abnormality, students with PWS do not register the feeling of fullness. There is varying degrees of food seeking. Many sneak and/or steal food – are at great risk for choking. Gain weight on ½ calories of other students; require calorie restricted diet & supervision around all food.	Receive/follow prescription from health care professional for calorie-restricted diet. Supervise student around all food sources. Keep food out of sight. Avoid use of food in classroom activities or as reward. Promptly empty garbage cans that contain discarded food. Train staff in the Heimlich maneuver. Have plan for how to handle food treats and other food issues in the classroom.	Severe Stomach Illness – Lack of Vomiting Severe stomach illness has been noted in students who have had a binge eating episode. Symptoms: abdominal bloating, vomiting, pain may or may not be present, general feeling of not feeling well. Rare for a person with PWS to vomit.	If symptoms of stomach illness are present, notify parent. Student should be urgently evaluated by a health care professional. Report any incidence of vomiting to the parent. Encourage the student to share honestly if they have had a binge episode. The student should not be punished if this has occurred.
Osteoporosis High risk due to hormone abnormalities & dietary limitations.	At high risk for fracture – assess injuries for possible sprain/fracture. May require x-ray to rule out fracture.	Increased Sensitivity to Medications More sensitive to medications that can cause sedation or sleepiness	Be aware of all medications that student is taking. Report any problems to parents.
Daytime Sleepiness Common to see in students. Often a symptom of sleep apnea. May be result of weak chest muscles-poor air exchange.	Physical therapy evaluation for muscle strengthening. Get student up and moving if fatigue is noted. May require a rest time during the school day. Communicate problem to parent & health care provider.	Scoliosis and Other Spine Problems Common to see scoliosis and other spine deformities in students’ w/PWS. Often difficult to detect if obese. May require bracing.	If found, refer to orthopedic specialist. Support and assist if brace is needed. Adaptive measures may be needed for physical education. Physical therapy evaluation for muscle strengthening.
Strabismus Often seen in younger students. Poor muscle tone/control in eyes Glasses, patching and in some cases surgery is needed.	Look for signs during vision screening. Refer to eye specialist if needed Make sure students wears glasses and/or patches if needed.	Dental Problems – Dry Mouth Common problems: o thick, sticky saliva, o teeth grinding, o rumination and cavities	Teach and encourage good dental care and water. Assist in referral to dentist if needed.

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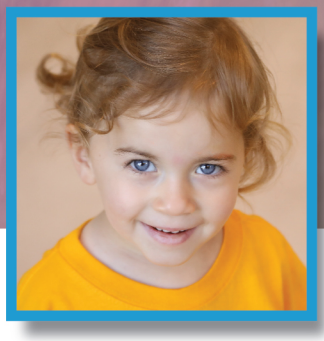
PWSA SCHOOL SUCCESS KIT
INDIVIDUALIZED EDUCATION PROGRAMS:

Medical Overview for School Nurse and Medical Support Staff



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Medical Overview for School Nurses and Medical Support Staff

Cause and Diagnosis of PWS

- **PWS occurs from three main genetic errors.** Approximately 70% of cases have a non-inherited deletion in the paternally contributed chromosome 15. Approximately 25% have maternal uniparental disomy (UPD) - two maternal chromosome 15s and no paternal chromosome 15. Additionally, 2-5% have an error in the “imprinting” process that renders the paternal contribution nonfunctional; rarely, these imprinting defects may be inherited.

- **Diagnostic testing** Individuals who have several the clinical findings should be referred for genetic testing. DNA methylation analysis confirms diagnosis of PWS. FISH and DNA techniques can identify the specific genetic cause and associated recurrence risk. Patients who had negative or inconclusive tests with older techniques should be retested.

Life Threatening Medical Concerns

- **Anesthesia, medication reactions:** Unusual reactions to standard dosages of medications and anesthetic agents may occur because of metabolic differences and obesity seen in PWS. A narrow airway may be present. Use extreme caution in giving medications that may cause sedation; prolonged and exaggerated responses have been reported. Several genes for GABA receptor subunits are located in the PWS chromosome region and are missing in patients with the deletion. This decrease in GABA receptors in PWS could alter the response to GABA receptor agonist sedative agents (propofol, benzodiazepines).
- **High pain threshold:** Lack of typical pain signals is common and may mask the presence of infection or injury. Someone with PWS may have difficulty localizing pain or not complain of pain until infection is severe. Parent/caregiver reports of subtle changes in condition or behavior should be investigated for medical cause.
- **Respiratory concerns:** Risk may be increased for respiratory difficulties. Obesity, hypotonia, weak chest muscles, and sleep apnea are among possible complicating factors. Sleep studies for central and/or obstructive sleep apnea and hypoventilation should be obtained.
- **Lack of vomiting;** Vomiting rarely occurs. Emetics may be ineffective, and repeated doses may cause toxicity. This characteristic is of concern considering hyperphagia and the possible ingestion of uncooked, spoiled, or otherwise unhealthful food items. The presence of vomiting may signal a life-threatening illness.
- **Body temperature abnormalities:** Idiopathic hyper- and hypothermia have been reported. Hyperthermia may occur during outdoor activities in warmer temperatures, minor illness and in procedures requiring anesthesia.

- **Severe gastric illness:** Abdominal distention or bloating, pain and vomiting may be signs of life-threatening gastric inflammation or necrosis, more common in PWS than in the general population. Rather than localized pain, there may be a general feeling of being un-well. If an individual with PWS has these symptoms, close observation is needed. A CAT scan of the abdomen and/or endoscopy may be necessary to determine degree of the problem and possible need for emergency surgery. Gastric rupture can also occur if the person with PWS after an episode of binge eating and engorgement. This must be considered a possibility if abdominal symptoms are present.
- **Central adrenal insufficiency:** Studies suggest an increased incidence of CAI in individuals with PWS. Measurement of cortisol levels during a significant illness and supplementation of cortisol may be indicated.
- **Skin lesions and bruises:** Skin picking is common in PWS, causing open sores. In some situations, skin and rectal picking can be severe. Individuals with PWS also tend to bruise easily. Appearance of such wounds and bruises may wrongly lead to suspicion of physical abuse.
- **Hyperphagia (excessive appetite):** Insatiable appetite may lead to life-threatening weight gain, which can be very rapid and occur even on a low-calorie diet. Individuals with PWS must always be supervised in all settings where food is accessible. Those who have normal weight have achieved this because of strict external control of their diet and food intake. Water intoxication has occurred in relation to use of certain medications with anti-diuretic effects, as well as from excess fluid intake alone, producing lower electrolytes.
- **Obesity-related problems:** include hypoventilation, hypertension, right-sided heart failure

Potential Characteristics

- Any infant with hypotonia should be tested for PWS. The following common characteristics raise suspicion of a diagnosis of PWS.
- Decreased fetal movement, infantile lethargy, weak cry
- Feeding problems and poor weight gain in infancy
- Excessive or rapid weight gain between 1 and 6 years of age; central obesity in the absence of intervention
- Distinctive facial features — dolichocephaly in infants, narrow face/bifrontal diameter, almond-shaped eyes, small appearing mouth with thin upper lip and down-turned corners of mouth
- Hypogonadism — genital hypoplasia, including undescended testes and small penis in males; delayed or incomplete gonadal maturation; and delayed pubertal signs after age 16, including scant or no menses in women
- Global developmental delay before age 6; mild to moderate cognitive disabilities or learning problems in older children
- Hyperphagia/food foraging/obsession with food
- Possible behavior problems — temper tantrums, obsessive/ compulsive behavior; oppositional, rigid, possessive, perseverating, but also sweet and loving
- Sleep disturbances especially daytime sleepiness and sleep apnea
- Short stature for genetic background by age 15 if untreated with growth hormone

- Hypopigmentation — fair skin and hair compared with family, primarily in deletion subtypes
- Small narrow hands and/or feet for height/age. Straight ulnar border
- Osteoporosis — can occur much earlier than usual and may cause fractures; ensure adequate calcium, vitamin D, and weight bearing exercise; bone density test recommended
- Diabetes mellitus, type II — secondary to obesity; responds well to weight loss; screen obese patients regularly
- Dental problems — may include soft tooth enamel, thick sticky saliva, poor oral hygiene, teeth grinding, and infrequently rumination. Special toothbrushes can improve hygiene. Products to increase saliva flow are helpful.
- Speech articulation defects and dyspraxia
- Strabismus — esotropia is common; requires early intervention, possible surgery
- Scoliosis — can occur unusually early; may be difficult to detect without X-ray, kyphosis is also common in teens and adults

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The Student with Prader-Willi Syndrome: Information for Transportation Personnel

A positive, successful education experience for every student begins the moment they step on the bus each morning. The ride to/from school often sets the tone for the student's attitude and outcome for a positive day. Transportation personnel are a very important, although often neglected, part of the education team. The purpose of this handout is to provide transportation personnel with an understanding of Prader-Willi Syndrome and the steps they can take to ensure a safe, enjoyable experience for everyone.

What is Prader-Willi Syndrome (PWS)?

PWS is a complex developmental disability that results from a defect on the 15th chromosome. Because of an abnormality in the brain called the hypothalamus, these students face challenges in learning, behavior, and appetite control. The message of fullness never reaches their brain, and they are almost always in search of food. The intensity of their food drive can vary but all students require support and understanding to manage their dangerous and life-threatening drive for food. Food security must be provided at all times when these students are being transported to and from school. Food security means that all food and drinks are out of sight, secured, and even locked.

What Can Transportation Personnel Do to Assist the Student with PWS?

The following acronym was developed by Latham Centers to enable transportation personnel to engage in safe travel practices.

D- Driver keeps all food secure
R- Review the expectations before you drive
I- Intervene immediately
V- Vehicle Safety/ Environmental Checklist
E- Expect a Challenge
S- Safety is the #1 priority
A- Ask the person how they are feeling
F- Find an activity
E- Enjoy each other's company

Transportation personnel can follow the tips below when implementing the above acronym.

- All food and drinks, including those of the drivers and staff, must be secured and out of sight at all times.
- The student's lunch should be given to the bus driver or bus aide when getting onto the bus. This will keep food secure and prevent the student with PWS from eating the food prior to arriving at school.
- Food should not be used to reward good behavior on the bus. Try using stickers, erasers, or even a high-five and verbal praise.
- Implement a no-food and drink policy for all persons on the bus-no exceptions!
- Review the safe behaviors that you want to see from the student while the bus is in motion.
- If negative behaviors occur on the bus, pull over and call for help.

- The bus should be cleared of all food, food remnants, food containers, drinks, drink cups, including food remnants that may be stuck to the floor (e.g., gum) prior to the student with PWS boarding the bus.
- Build a friendly relationship with the students by engaging them in conversation upon arrival on the bus and throughout the ride, as appropriate. Know the student's name and a little about their daily lives and interests. [Note: Avoid teasing, sarcasm, and abstract humor as it is often misunderstood. On the other hand, feel free to make fun of yourself as you will surely get a smile and laugh].
- Give concrete answers to questions from the student (e.g., *Student:* Why is my aide not on the bus?
  Driver: Your aide was not feeling well today.
- Assign the student to an important job on the bus. For example, the student may be responsible for taking attendance for all the students as they board and get off the bus each day. This will keep the student with PWS engaged in a positive and meaningful activity.

Transportation personnel are an important part of the education team that helps support the student with PWS. For additional information about PWS, please call the Prader-Willi Syndrome Association | USA (PWSA | USA) at 1-800-926-4797 or visit www.pwsausa.org.

PRADER-WILLI SYNDROME

MEDICAL ALERTS

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PRADER-WILLI SYNDROME

Prader-Willi syndrome (PWS) is a complex neurobehavioral genetic disorder resulting from abnormality on the 15th chromosome. It occurs in males and females equally and in all races. Prevalence estimates range from 1:12,000 to 1:15,000. Incidence in newborns is unknown.

PWS typically causes low muscle tone, short stature if not treated with growth hormone, cognitive deficits, incomplete sexual development, problem behaviors, and a chronic feeling of hunger that, coupled with a metabolism that utilizes drastically fewer calories than normal, can lead to excessive eating and life-threatening obesity.

It is felt to be a multistage disorder with decreased fetal movement prenatally and low birth weight. Infants have failure to thrive due to feeding problems and hypotonia. Toddlers have increased weight gain, then hyperphagia and obesity as they get older, if calories are not restricted. Most of the medical problems in Prader-Willi syndrome are related to the obesity, hypotonia, and hypothalamic dysfunction.

Some of the other factors that may cause difficulties include adverse reactions to medications, high pain tolerance, gastro-intestinal and respiratory issues, lack of vomiting, and unstable temperature. Adrenal insufficiency may also occur.

Severe medical complications can develop rapidly in individuals with PWS.

Members of the Clinical Advisory Board are available for consultation with physicians through the Prader-Willi Syndrome Association | USA.

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Mission

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

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MEDICAL ALERT

Important Considerations for Routine or Emergency Treatment

Obesity and its related complications is the major cause of morbidity and mortality in Prader-Willi syndrome. Keeping the individual at a healthy weight will minimize these complications but there are important medical and behavior problems unique to Prader-Willi syndrome regardless of weight status.

Medical professionals can contact PWSA | USA to obtain more information and be put in touch with a specialist as needed. Up-to-date has an excellent summary of the syndrome.

Central Adrenal Insufficiency in Individuals with Prader-Willi Syndrome

Several studies have shown CAI in individuals with PWS while others failed to show a deficiency. Stress dose of cortisol may be indicated if individual has problems after surgery or during times of stress. **http://www.pwsausa.org** and view Medical section under Adrenal Insufficiency.

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, or decreased movement of the extremity for more than a few days may warrant an x-ray.

Hyperphagia (Excessive Appetite)

Individuals with PWS must be constantly supervised in all settings to prevent access to food. In hospital settings, obtaining unguarded food can lead to rapid ingestion and fatal choking. 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 no treatments for this relentless hunger. Insatiable appetite may lead to life-threatening weight gain, which can be very rapid and occur even on a low-calorie diet.

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.

Pain Insensitivity

Lack of typical pain signals is common and may mask the presence of infection or injury. Someone with PWS may not complain of pain until infection is

severe or may have difficulty localizing pain. Parent/caregiver reports of subtle changes in condition or behavior should be investigated for medical cause. Any complaint of pain by a person with PWS should be taken seriously.

Skin Lesions and Bruises

Because of a habit that is common in PWS, open sores caused by skin picking may be apparent. Individuals with PWS also tend to bruise easily. These lesions can cause serious life-threatening infections. There are approaches to help mitigate picking. **h p://www.pwsausa.org/resources/medical-issues-a-z/** and view skin picking. Appearance of such wounds and bruises may erroneously lead to suspicion of physical abuse.

Swallowing and Choking

Persons with PWS are highly likely to have an undetected swallowing problem that places them at risk for asphyxiation of a food bolus (choking), and they require a specific type of swallowing evaluation. A clinical or bedside evaluation is not sufficient to detect dysphagia in this population. They frequently cannot tell if they cleared their airway after swallowing, increasing the risk for aspiration. Choking can also occur with rapid ingestion of unguarded foods and has led to many deaths in the PWS population.

http://www.pwsausa.org/resources/medical-issues-a-z/ and view Choking/Swallowing.

Temperature Abnormalities

Idiopathic hyper- and hypothermia have been reported. Hyperthermia may occur during minor illness and in procedures requiring anesthesia. Fever may be absent despite serious infection. All individuals with PWS are at risk for mild hypothermia because of impaired peripheral somatosensory and central thermoregulation, poor judgment and cognitive inflexibility. Malignant hypothermia is a life-threatening problem occasionally seen in PWS. **http://www.pwsausa.org/resouces/medical-issues-a-z/** and view Temperature.

Vomiting – Lack of ability to vomit

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

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. Anti-diarrheal medications may cause severe colonic distension, necrosis and rupture and should be avoided.
http://www.pwsausa.org/resources/medical-issues-a-z/ and view Water Intoxication.

Respiratory Concerns

Individuals with PWS are at increased risk for respiratory difficulties. Hypotonia, weak chest muscles, swallowing abnormalities and sleep apnea are common. Anyone with significant snoring, regardless of age, should have a medical evaluation to look for obstructive sleep apnea. Infants commonly have central sleep apnea which generally improves over time but may also have obstructive sleep apnea due to hypotonia and other factors. Hypotonia can lead to diminished activity levels and low aerobic capacity. Hypoventilation may be central in origin.
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 apnea or obesity are at more risk for reflux.

Recommendations for Evaluation of Breathing Abnormalities Associated with Sleep in Prader-Willi Syndrome

PWSA | USA Clinical Advisory Board Consensus Statement - 12/2003

Problems with sleep and sleep disordered breathing have been long known to affect individuals with Prader-Willi syndrome (PWS). The problems have been frequently diagnosed as sleep apnea (obstructive [OSA], central or mixed) or hypoventilation with hypoxia. Disturbances in sleep architecture (delayed sleep onset, frequent arousals and increased time of wakefulness after sleep onset) are also frequently common. Although prior studies have shown that many patients with PWS have relatively mild abnormalities in ventilation during sleep, it has been known for some time that certain individuals may experience severe obstructive events that may be unpredictable.

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 hypoxia. Due to a few recent 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 hypoventilation or OSA. 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 new 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, upper airway obstruction, 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 pulmonary specialist knowledgeable about treating sleep disturbances to ensure that a detailed plan for treatment and management is made. Referral to a pediatric or adult pulmonologist with experience in treating sleep apnea 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 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 training 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 (IBW > 200%) 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 persons experienced with PWS.

If airway related surgery is considered, the treating surgeon and anesthesiologist should be knowledgeable about the unique pre- and postoperative 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.

Growth Hormone Treatment and Prader-Willi Syndrome

PWSA | USA Clinical Advisory Board Consensus Saemen - 6/2009
PWSA Inernational Consensus Saemen 2013

Both statements are found at **http://www.pwsausa.org/resources/medical-issues-a-z/** and view Growth Hormone.

In-Patient Considerations

Access difficulties – venous and airway

Obesity and poor muscle tone may complicate line placement. A small airway, high palate, and/or obesity may complicate ability to intubate. Saliva is often thick and sticky. Many persons with PWS will have difficult IV access due to increased fat mass and smaller than normal blood vessels. Outpatient procedures and general sedation may be especially problematic. Care must be taken during procedures done in out of hospital settings, and that proper equipment for resuscitation is immediately available and consideration for doing these procedures in an OR should be discussed. Procedures where more than light sedation is used may warrant an overnight observation.

Anesthesia

People with PWS may have unusual reactions to standard dosages of anesthetic agents. Use caution in giving anesthesia. Serious problems occur during conscious sedation, if it is not well monitored, rather than from the use of general anesthesia and airway management. Ongoing assessment of breathing and oxygen saturation is critical in all outpatient procedures including dental work.

http://www.pwsausa.org/resources/medical-issues-a-z/ and view Anesthesia.

- Anesthesia and Prader-Willi Syndrome: James Loker, M.D., Laurence Rosenfield, M.D.
- Anesthesia Concerns for Patients with PWS: Winthrop University

Behavior problems

Individuals are prone to emotional outbursts, obsessive-compulsive behaviors, and psychosis. Psychotropic medications may affect metabolism of anesthesia leading to shorter or longer duration of action.

Cardiac problems

Surprisingly, coronary disease is less in PWS than in individuals with similar obesity. Cardiac problems usually are due to hypoventilation right heart failure. Edema can often be seen in the obese individual

even in the absence of heart failure and is treated by weight loss and ambulation. Diuretics are usually not that beneficial in treating the edema.

Food seeking behaviors/Relentless hunger

Complete safety from access to food is essential in any health care setting. Assume individual has eaten unless verified by 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 caloric restricted diet and that should be conveyed to the nutritionist and kitchen.

Hypothalamic dysfunction – Pituitary deficiencies

Hypothyroidism- Risk of central (TSH deficiency) hypothyroidism is 20-30% and may be undiagnosed prior to surgery.

Growth hormone deficiency - All individuals should be considered to be GH deficient.

Hypothalamic dysfunction is also the presumed origin of many other unique problems including temperature regulation, hunger, ventilatory effort, behavior patterns.

Hypotonia

This muscle weakness may complicate ability to cough effectively and clear airways.

Narcotics

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

Obesity

Consideration for obstructive apnea, pulmonary hypertension, diabetes, and right heart failure should be addressed.

Pain insensitivity

Unexplained tachypnea or tachycardia may be the only indication of pain. Behavior problems which are not typical for this person may be evidence of pain. Individuals with PWS may not respond to pain in the same manner as others and it may mask the presence of underlying problems. Since pain may not be present, other signs of underlying problems should be monitored.

Pulmonary embolism

Individuals with PWS are at risk for pulmonary embolism. DVT prophylaxis should be considered in all obese individuals. Prolonged bed rest is to be avoided.

Psychosis

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

Saliva abnormalities

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

Skin picking

May complicate healing of IV sites and incisional wounds. Restraints or gloves may be necessary to protect wounds during healing.

Temperature instability

Low basal temperature is typical in healthy individuals with PWS. Hypothalamic dysregulation can lead to poor control during fever or hypothermia. There is no known predisposition to malignant hyperthermia, but depolarizing muscle relaxants should be avoided if possible.

Surgical and Orthopedic Concerns

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

Musculoskeletal manifestations, including scoliosis, hip dysplasia, fractured bones (which may be undetected), osteoporosis and lower limb alignment abnormalities, are described in the orthopedic literature. However, care of this patient population from the orthopedic surgeon's perspective is

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

Postoperative Monitoring of Patients with Prader-Willi Syndrome

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

- Abnormal physiological response to hypercapnia and hypoxia
- 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 postoperative treatment procedures due to:
  - Extreme food seeking behavior and hyperphagia due to hypothalamic dysfunction
  - High incidence of gastroparesis and slow motility of the intestinal tract

– Extreme skin picking which may interfere with wound healing
– Altered temperature regulation – fever may be absent in the presence of infection. There does not seem to be a higher incidence of malignant hyperthermia
– The possibility of central adrenal insufficiency

RECOMMENDATIONS:

- 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 postoperative with attention to airway and breathing.
- A conservative approach to pain management and use of narcotic agents.
- Full assessment of return of GI motility prior to initiation of intake by mouth 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 postoperatively.

- 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.
- Utilization of respiratory therapy interventions to prevent atelectasis and/or postoperative 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.

http://www.pwsausa.org/resources/medical-issues-a-z/ and view Postoperative Monitoring.

Severe Gastric Intestinal Concerns

Vomiting – Lack of ability to vomit

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

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. 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 monitoring, 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.

In addition to gastric distension, colonic impaction may also be present and need to be addressed. 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.

Constipation in Individuals with Prader-Willi Syndrome

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Constipation is a common problem in individuals with Prader-Willi syndrome (PWS). It takes longer for food to move through the GI system 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. Failure of standard constipation protocols to clear the stool in a timely manner, especially in the face of increasing abdominal distension, vomiting, decreased appetite, stoppage of food consumption and/or abdominal pain warrants surgical or GI consultation. Emergent surgical or colonoscopic intervention may be necessary.

Kuhlmann, et al. (2014) A descriptive study of colorectal function in adults with Prader-Willi syndrome: high prevalence of constipation.* ***BMC Gastroenterology*, Apr 4; Vol 14: page 63

Prader-Willi Syndrome | USA ALERT! Risk of Stomach Necrosis and Rupture

Possibly Related to Chronic Gastroparesis

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

Signs and symptoms of stomach necrosis and rupture:

- **Vomiting-** Any vomiting is unusual in Prader-Willi syndrome
- **Loss of appetite-** (ominous sign)
- **Lethargy**

- **Complaints of pain, usually non-specific-** Pain sensation is abnormal in Prader-Willi syndrome due to high pain threshold; 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**
- **Temperature regulation** is altered in Prader-Willi syndrome
- **Guaiac positive stools (chronic gastritis)**

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

These Signs should raise suspicion of STOMACH NECROSIS/RUPTURE as a possible diagnosis which can be LIFE-THREATENING!

History may include:

- **History of binge eating within the week**
- **Hyperphagia and binge eating are characteristic** of Prader-Willi syndrome, regardless of whether obese or slim

- **Frequently occurs** after holiday, or social occasion with less supervision of intake
- **History of gastroparesis-** Common in Prader-Willi syndrome, though often undiagnosed
- **Often slim or history of significant obesity followed by weight loss-** May leave the stomach wall thinned
  http://www.pwsausa.org and view Medical A-Z under GI Problems.

IN THE EVENT OF DEATH

In the case of a death or impending death, please call PWSA |USA immediately **at 1-800-926-4797** for support and advice.

Reporting of Deaths

The Prader-Willi Syndrome Association | USA has created a research database of reported deaths of individuals with PWS. Although most premature deaths are attributable to morbid obesity, cases unrelated to obesity have been noted. PWSA | USA has a formal investigation of causes of death.

PWSA | USA also provides bereavement support to families who have lost a child with PWS. Please call PWSA | USA to report a death and so the family can receive grief counseling.

Organ Donation for Research

When a child or adult with PWS dies, the family may wish to consider donation of organs for research. PWSA | USA has established a procedure for such donations.

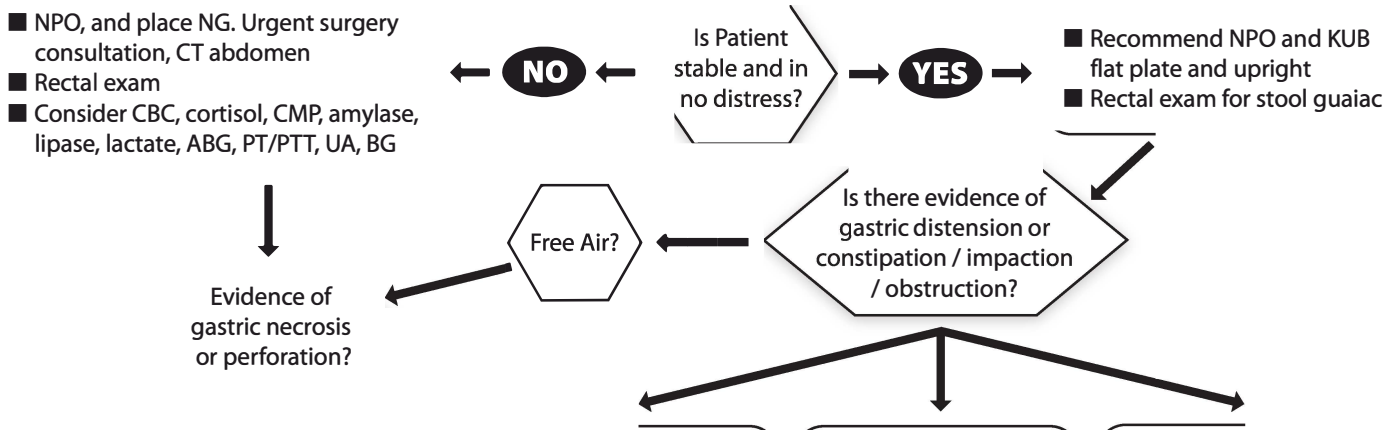
For donating brain tissue, contact Autism BrainNet 877-333-0999.

http://www.pwsausa.org and view Brain Tissue Donation.

Important Notes

Evaluation of Individuals with Prader-Willi Syndrome with GI Complaints

Known or suspected binge, vomiting, lethargy, or abdominal pain/distension
Admit Patient for Observation



YES

Emergent Surgery
There is no advantage to waiting for surgery.

NO

- Admit to ICU for observation
- Surgical consultation
- Check plain film every 6–8 hours unless examination changes
- Assess for other etiologies

Constipation

- Stool cleanout regimen
- Check for impaction either digitally or with fleet enema prior to starting oral cleanout
- Calorie restricted clear liquid diet
- Check TFTs, electrolytes
- Consider GI or surgical consultation, especially if stool guaiac positive
- Reassess plain film in 12-24 hours
- Increasing distension or failure to clear stool may indicate impaction or obstruction. Further GI consultation should be considered

Gastric distension

- NPO
- NG for decompression
- Make surgery aware of Patient
- Reassess plain film in 6-8 hours unless condition worsens then urgent CT
- Consider GI consultation, especially if guaiac positive stool

Neither

- Check for UTI, pancreatitis, cholelithiasis, etc.
- RUQ ultrasound
- If patient vomiting consider esophageal obstruction or toxic ingestion

NOTE:
Individuals with Prader-Willi syndrome have decreased muscle tone and pain response so clinical evaluation for acute abdomen is altered similar to a patient on steroids.

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

Credits: James Loker, M.D., Pediatric Cardiologist • Ann Scheimann, M.D., M.B.A., Gastroenterologist PWSA|USA Clinical Advisory Board Members

www.pwsausa.org

Important Notes

Prader-Willi Syndrome Medical Alerts
by
Medical Specialists in Prader-Willi Syndrome

This life-saving Medical Alerts Booklet is dedicated to
Janalee Heinemann in apprecia ion for a life ime 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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