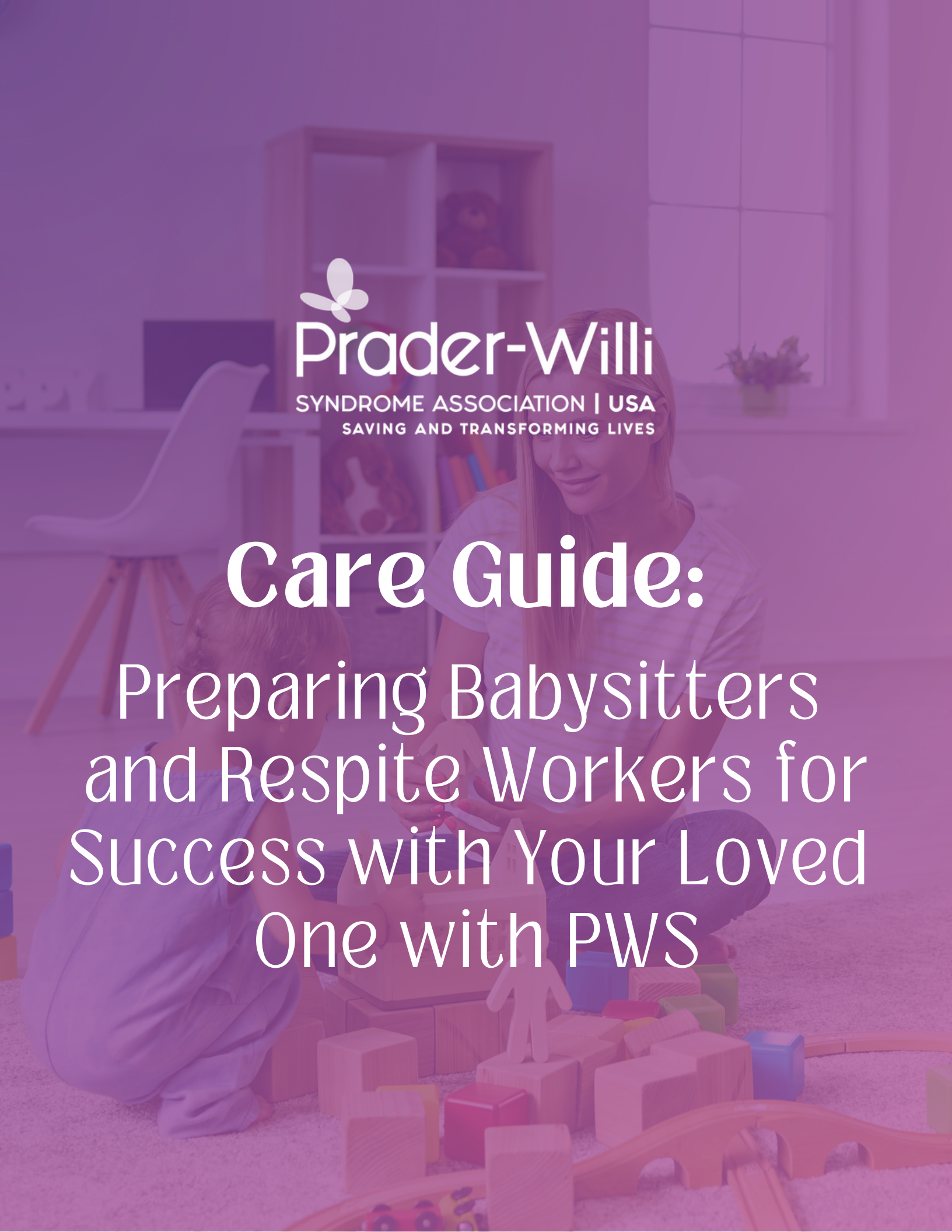

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

Care Guide:

Preparing Babysitters and Respite Workers for Success with Your Loved One with PWS

Welcome to Your Care Guide

Created by Prader-Willi Syndrome Association | USA

As a parent / caregiver of someone with Prader-Willi syndrome, you shoulder a tremendous amount of love, responsibility, and care each day. We know that entrusting your loved one to a babysitter or respite worker can be scary, but having the right tools in place can make all the difference.

This ***Care Guide*** was designed to support you in preparing babysitters and respite workers with the essential knowledge they need to care for your loved one with PWS. From documenting medical details and daily routines to outlining favorite activities and areas where assistance is required, this guide ensures that caregivers have a comprehensive understanding of your loved one's unique needs.

We also recognize the importance of taking time for yourself. Whether it's running errands, enjoying a date night, or simply recharging, you deserve peace of mind knowing that your loved one is in capable, well-informed hands. This guide helps provide that assurance while enabling you to step away when needed, knowing your child's care is well-managed.

Included in this toolkit are two **fillable forms** to document essential information, as well as **helpful resources** like "Supporting Someone with PWS" and the "PWS Medical Alerts Booklet."

Should any questions arise or additional support be needed, PWSA | USA is here for you with a 24-hour emergency crisis line at 941-312-0400.

Your dedication and care are the foundation of your loved one's success, and we're honored to provide resources to help lighten the load.

Important Information for Babysitter or Respite Worker

Prader-Willi SYNDROME ASSOCIATION | USA
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Our loved one has Prader-Willi Syndrome. Here are some important things to know.

Address (for 911): _____

Main Emergency Contact(s): _____

_____

Pediatrician Name/Phone #: _____

Alternate Contacts: _____

_____

Secondary Diagnosis: _____

Medications/ Supplements: _____

_____

WiFi Password: _____

Poison Control #: _____

Other important Info: _____

_____

_____

PWSA | USA Crisis Phone Line: 941-312-0400

Website to find additional resources: www.pwsausa.org

About Our Loved One

Prader-Willi SYNDROME ASSOCIATION | USA
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Details of Diagnoses

- Prader-Willi Syndrome:
  - See attached brochures
  - Food security is very important! No food is to be given other than what has been previously discussed or set aside. They must be fully supervised while eating or while the food is accessible.
- Secondary Diagnosis *(if there is one)*: _____
  - Important details: _____

_____

_____

Interests (e.g., movies, books, characters, games, activities)

_____

_____

_____

Assistance Needed (e.g., use timer or countdown when transitioning between activities, offer only 2 options when choosing a book or game, must be supervised when outside)

_____

_____

_____

Things We're Working On (e.g., saying please and thank you, washing hands after using the bathroom)

_____

_____

_____

Stimming Behaviors / Signs of Anxiousness - How to Manage (e.g., skin picking - distract, find something to occupy hands; perseveration on topic - give specified amount of time to discuss and move on)

_____

_____

_____

What is Prader-Willi Syndrome?

Prader-Willi syndrome is a disorder of chromosome 15 Prevalence: 1 : 12,000-15,000 (both sexes, all races) .Major characteristics: hypotonia, hypogonadism, hyperphagia, cognitive impairment, challenging behaviors. Major medical concern: morbid obesity

Food and Weight Challenges

Prader-Willi syndrome is best known for its symptom called "hyperphagia" which means literally "over eating." For reasons not yet understood, the brain of someone with PWS drives the individual to want to eat as much food as possible all of the time. Persons with PWS feel a constant drive to eat that is never satisfied no matter how much food is eaten. At the same time, metabolism rate slows to about half what it should be. If caloric intake is not significantly reduced and strictly controlled, persons with PWS will quickly gain an enormous amount of weight leading to complications caused by morbid obesity. People with PWS are vulnerable to sudden death due to choking, stomach rupture, or stomach necrosis.

Just because someone has PWS doesn't mean they are destined to become obese. People with PWS can absolutely maintain a healthy weight, but it takes constant and continuous supervision from not only their parents or primary care providers, but from everyone in whose care they are entrusted.

Behavioral Challenges

Second *only* to hyperphagia on the list of PWS challenges is the misbehavior that many people with PWS exhibit.

The brain of someone with PWS causes individuals to feel a global sense of anxiety, think oppositionally, have a low tolerance to frustration, be argumentative, and react to disappointment and frustration with tears or anger. Regardless of age, persons with PWS can exhibit temper tantrums that can include yelling, foul language, hitting, kicking, sitting and other refusal behaviors.

- This rare medical disorder, pronounced PRAH-der WILL-ee, affects multiple systems in the body and causes many different symptoms. Many of these symptoms are very serious and some of them are life-threatening.
- People with Prader-Willi syndrome (PWS) experience a host of challenges including weak muscles, problems with balance and coordination, developmental delays, speech and language problems, temperature regulation problems, orthopedic problems, and sleep problems, to name a few.
- This brochure will provide you with an overview of important information you need to know when someone with Prader-Willi syndrome is in your care.

Prader-Willi Syndrome Association | USA has an extensive amount of material for parents, extended family, care providers, physicians, therapists, caseworkers, and school staff. Contact PWSA | USA to learn more about Prader-Willi syndrome and to receive support and advocacy services for you, your child, your friend, patient, or student.

Prader-Willi Syndrome Association| USA, established in 1975, is an organization of families and professionals working together to raise awareness, offer support, provide education and advocacy, and promote and fund research to enhance the quality of life of those affected by Prader-Willi syndrome.

Prader-Willi Syndrome Association | USA

1032 E Brandon Blvd, #4744
Brandon, FL 33511

Tel: 941.312.0400
E-mail: info@pwsausa.org
Web: www.pwsausa.org

**PWSA | USA, a 501(c)(3) non-profit organization, is very grateful to our donors and invites support of our organization. All contributions are tax deductible to the extent allowed by law.*

We hope you find these materials helpful and that you consider a donation to PWSA | USA to assist in developing more good work(s) like this. Please see our website, www.pwsausa.org .

Supporting Someone with PWS

Whether you are a playmate or classmate, neighbor, relative or friend, babysitter or companion; it helps to know about Prader-Willi syndrome.

Prader-Willi
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It's important to understand how persons with PWS typically think and react so that you can better avoid the behavioral problems that are common in persons with PWS.

Strategies to Manage Symptoms

There are currently no known medications that eliminate or even reduce the insatiable appetite that is the hallmark symptom of PWS. There are, however, therapeutic and environmental interventions that can help manage weight, improve behavior, save lives, and improve the quality of life of all who are impacted by Prader-Willi syndrome.

Food Security

People with PWS cannot manage their own food intake because their brain tells them to eat as much food as possible, which means they must depend upon everyone in their environment to make sure they are safe around food. ***The Principles of Food Security,*** a concept coined by PWS specialists Linda Gourash, MD and Janice Forster, MD, describe the basics of how to manage the food environment:

- **No Doubt** that appropriate foods will be served at the appropriate time **plus No Hope** that extra, unauthorized foods can be obtained ***equals* No Disappointment.** No disappointment means fewer behavioral problems.
- People with PWS need to know **that they will eat and when** they will eat. Having a written schedule when meals and snacks will be served is very helpful because once it's written, it's often not argued about. Stick to the schedule as persons with PWS cannot be expected to be flexible.
- It's helpful for the person with PWS to eat every 2 1/2 to 3 hours. Eating this frequently, however, requires a daily diet **very low** in calories, fat, and carbohydrates. Some people with PWS cannot exceed 800 calories per day.
- **NEVER use food as a reward or punishment.** For someone with PWS food must be as stable as oxygen, **never** contingent upon good behavior and **never** taken away for poor behavior.
- **It is imperative to follow the menu instructions *EXACTLY AS DICTATED* by the parent.** Because the metabolic rate is very slow, every calorie consumed by someone with PWS can potentially cause weight gain. Giving someone with PWS extra food causes not only weight gain but gives them hope that they will receive extra food from you later, and that hope will likely lead to a behavior problem some time down the road. There are many ways other than food to provide a special treat or show you care, such as, stickers, small toys, time playing together, employing the individual as your special helper, and hugs.
- **Secure and lock up all food sources.** Almost all families who live with someone with PWS lock their refrigerator and food pantry. Not only does locking away food keep the individual with PWS safe, it tells their brain that food is not available and reduces the hopeful anxiety they feel to obtain that food. In addition, secure wallets, purses, and loose change because children and adults with PWS know that money can purchase food. Stealing is a symptom of PWS.

You will show the person with PWS and their family that you care about them by securing all food sources and by following all parental instructions.

Behavioral Interventions

It's important to familiarize yourself with how PWS typically impacts the brain so you can minimize or avoid behavior problems. In general, work to create an environment that is predictable, positive, and free of stress. Use positive words, praise and encouragement; use a calm tone of voice; use rewards such as your attention, stickers or small toys; and engage the individual as your 'special helper.'

- **Anxiety** Almost all persons with PWS feel an underlying degree of anxiety all of the time and do best when they know what to expect and what is expected of them. Create a written schedule of the day's activities including approximate timing of all snacks and meals. "Paint a picture" of new activities or situations; talk about what things might look like, sound like, what can be expected to happen, etc. Talk about your expectations of behavior, and make a plan for what will happen if there is a behavior problem. Stick to the schedule to the extent possible as persons with PWS cannot be expected to be flexible.
- **Low Tolerance to Frustration** Almost all people with PWS have difficulty managing feelings of frustration and disappointment. To the extent possible, eliminate the potential for frustrators and disappointment and use positive-toned language. For example, the person with PWS says, "I want to go to the movies" but you know you can't schedule it today. Instead of saying, "No, we can't do that today" reply, "That sounds great! I'd like to go to the movies too!Today we need to xyz. Let's see what movies are playing and plan to go tomorrow!"
- **Hyper-Reactive** Almost all people with PWS can be described as "hyper-reactive," meaning they tend to become overly upset about a seemingly minor incident. When someone with PWS becomes upset, it's best not try to talk them out of their upset as this will cause them to become even more upset and work even harder to show you how upset they are. Instead, immediately and calmly demonstrate understanding and empathy as this will help them recover more quickly. For example, "You want to see a movie today. You don't want to wait. I'm so sorry." Then stop talking or only occasionally and calmly repeat their upset. People with PWS are often concerned with "fairness." Sometimes simply agreeing to show you understand their upset will be calming, "You're right, it's not fair. I'm sorry."
- **Oppositional** Most people with PWS are oppositional; the brain reflexively thinks, "No!" Strategies to counter this are to refrain from giving "yes or no" choices. For example, instead of asking, "Are you ready to get dressed now?" give two or three preferred choices such as, "Do you want to get dressed in 3 minutes or 5 minutes?" As much control over things the individual with PWS perceives he or she has, the better will be their behavior.
- **Inflexible** People with PWS often have a difficult time transitioning from one thought or activity to the next. Give prompts to end a conversation about a particular topic: "This has been fun to talk about xyz. You can say one more thing and then we will talk about something else." Give incremental prompts before moving to the next activity. For example prompt, "Five minutes before we need to leave... Ok, 3 minutes before we need to leave so start putting away your books... Ok, 1 minute. Everything should be put away. Get your shoes on." Some people with PWS move more slowly so make sure you carve out lots of time for transitions.
- **Never give in to a temper tantrum or a sit down strike whether it's for food or anything.** Once you give in, you have taught the person with PWS that next time you will eventually give in if they just cry louder, longer, or behave with increased aggression.
- **Have patience.** And then have more patience. People with PWS typically process or think about things a bit more slowly and may need extra time to comply with a request.
- **Have fun!** When the needs of the individual with PWS are addressed, there will be few, if any, behavioral problems. So enjoy the time you spend in the company of your loved one or friend with Prader-Willi syndrome!

Special Health Concerns:

High Pain Threshold Diminished pain response. All injuries and illnesses must be carefully evaluated.

Altered Temperature Regulation Little or no fever present with illness. Low tolerance to external heat or cold temperatures.

Severe Stomach Illness Life-threatening situation often seen after binge episode. Abdominal bloating and/or pain, vomiting, a general feeling of illness. Must urgently seek medical care.

Choking Often eat very fast. If successful in stealing food, will ingest food quickly and risk choking.

Skin Picking Common to see open sores; may pick at various openings of body.

Excessive Daytime Sleepiness Low endurance. Sleep apnea and hypoventilation common. Some require rest periods; others may need increase in activity.

Strabismus Cross-eye often detected in visual screenings. May require referral to eye specialist for patching and/or surgery.

Scoliosis May require bracing and/or surgery if severe.

Each student's individual needs must be closely evaluated, and an educational plan developed.

What is Prader-Willi Syndrome?

PRADER-WILLI SYNDROME (PWS) is a complex developmental disability that results from a defect on the 15th chromosome. It causes a malfunction in the area of the brain called the hypothalamus.

Common Characteristics:

Hypotonia (low muscle tone), Low Stamina, Developmental Delays, Cognitive or Learning Disabilities, Speech and Language Delays/ Problems, Behavior Challenges, Complex Health Issues and the Inability to Control his/her Appetite

Students with PWS require services and support from many educational professionals. Most qualify for the following related services:

Early Intervention Assessment and Services Birth to 3 and Early Childhood services address motor, speech and developmental delays.

Speech and Language Varying degrees of speech and language problems.

Occupational and Physical Therapy Fine and gross motor weakness, trunk/core muscle weakness which impacts endurance, breathing and spine problems.

Psychological and Counseling Services Cognitive assessments and behavior management issues.

School Health Services Unique health needs and issues require ongoing assessment and oversight.

Parent Counseling and Training Help in management of global learning, behavior and health issues.

Transportation Safety, hypotonia, behavior, cognitive, and food seeking issues often necessitate services.

Prader-Willi Syndrome is:

A non-hereditary birth defect resulting from a disorder of chromosome 15

A serious, life-long, and life-threatening medical condition

Occurs in 1:12,000 to 1:15,000 births, both sexes, all races

Characterized by
- Hypotonia *(low tone)*
- Hypogonadism *(underdeveloped sex organs)*
- Hyperphagia *(uncontrollable hunger)*
- Cognitive impairment
- Challenging behaviors

One of the most common conditions seen in genetic clinics

The most commonly known genetic cause of life-threatening obesity

A major medical concern is morbid obesity, however with early diagnosis and early intervention, many children can maintain a healthy weight.

For more information on how to successfully support the student with PWS, contact:

Prader-Willi Syndrome Association | USA

1032 E Brandon Blvd, #4744
Brandon, FL 33511

Tel: 941.312.0400
E-mail: info@pwsausa.org
Web: www.pwsausa.org

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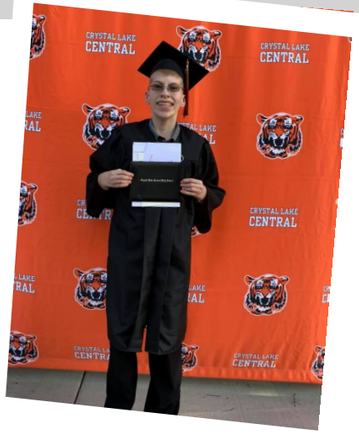
What Educators Should Know



Students with PWS are very caring, loving, sensitive and conscientious. They want very much to be successful, have friends and be a part of their school community.

Prader-Willi
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Although faced with some unique challenges, students with PWS can play, learn, work and live successful in our communities.

A team of knowledgeable educational staff help to make this possible.

Common Issues and/or Behavior Challenges that Impact Learning:

Strong Food Seeking Behavior

The message of fullness never reaches their brain.

Can gain weight on ½ calories of others their age. Must be on low calorie diet. Can become irrational when it comes to food-related issues. May steal or fight to obtain food.

Recommendations:

Create a "food-free" learning environment. Remove food from sight.

Avoid using food as a manipulative, incentive, or reward.

Supervise in all areas where food or money can be accessed – lunchroom, hallways, break rooms, vending machines, offices, and special events.

Follow calorie restricted diet as prescribed by a health care professional.

Work with parents in developing a plan to handle birthday and other special events where treats may be present.

Create a positive behavior support plan that emphasizes appropriate, supported behavior in the event that the student steals food or money.

- Encourage "good choices" – avoid trading or sharing; practice returning item.
- Avoid threats.
- Do NOT use physical force to take it away.
- Communicate with parents so calories can be adjusted.

Monitor weight. Include exercise.

Tenuous Emotional Control

Can lose control of emotions and become over-stimulated easily.

Have knowledge and skill deficit in handling frustration and anger.

May yell, cry, swear, destroy property and/or do self-injury.

Recommendations:

Be aware of over-stimulating situations – especially in hallways. Carefully plan transitions, arrivals and dismissals.

Develop a behavior support plan that includes teaching the student what **to do** if he/she feels angry or frustrated.

Encourage communication of feelings – using words. Listen to what is said.

Don't try reasoning if out of control. Limit discussion.

Rigid, Perseverative Thinking

Common to receive and store information in very orderly manner. Strong need for structure & routine.

Do not handle changes well.

Often want to finish what is started. Transitioning to new things is difficult.

Can get caught on one idea or issue to the point where it overshadows learning. Can lead to loss of emotional control.

Recommendations:

Foreshadow schedule for the day. Put any changes in writing.

Keep instructions simple.

Provide praise when being flexible.

Use reflection – have student restate what was just said.

Limit repetitions – by number or time.

Less is best – give fewer amounts to do and "add on" for extra credit.

Elopement

Some students with PWS like to run away when faced with situations they feel they cannot handle.

Recommendations:

Teach, practice, and encourage other ways for the student to handle feelings associated with these situations.

Provide close supervision.

Incentive programs for positive behavior.

Difficulty with Peer Interactions

Many are very friendly and outgoing but lack age-appropriate social skills.

Can get caught up on the idea of "fairness" and compare themselves to others.

Want friends and to be able to participate in social and extra- curricular events.

Recommendations:

Include in social skill classes and coaching to assist in developing appropriate skills – sharing, taking turns, losing appropriately.

Have the student role play and practice different social situations.

Create supported play groups, recess, and other social outings.

Clearly communicate "dos and don'ts" for dating and social interactions. Visual format helpful.

Provide support and modifications so the student can participate in social and extra-curricular events.

Learn the Student's Strengths:

Most love to be "helpers". Put them in leadership roles where they can succeed.

Many are visual learners. Put things in writing or in another visual format.

Some are very social. Teach appropriate times and ways for them to maximize this skill.

Build up their self-esteem. Most love rewards and awards. Certificates and other acknowledgments can be a great way to celebrate success.

Practice during times where there is no stress. Use their obsessive-compulsive tendency to help them learn and develop more appropriate strategies.

Talk with the parents to learn other areas of interests and strengths.

Communication is Key!

It is very important to keep the lines of communication open when educating and working with the student with PWS and his/her family. This can be done in a number of different ways – e-mail, voice mail, notebooks and/or in- person.

Report and celebrate successes. Share important events and/or assignments. Problem solve around issues ahead of time. Identify concerns and, if needed, set up monthly meetings to keep the lines of communication open.

Look for our other great PWSA (USA) resource titled:

How Does a Person with PWS Think?

Quality of Life Issues

General health is usually good in individuals with PWS. If weight is controlled, life expectancy may be normal, and the individual's health and functioning can be maximized. The constant need for food restriction and behavior management may be stressful for family members. PWSA | USA can provide information and support.

Adolescents and adults with PWS can function well in group and supported living programs, if the necessary diet control and structured environment are provided.

To date, no medication or surgical intervention has been found that would eliminate the need for strict dieting and supervision around food. Bariatric procedures do not address the central lack of satiety and put the individual at risk for complications.

Studies show improvement in linear growth, fat mass, motor strength, respiratory drive, and bone density with the use of **growth hormone** in PWS. Precautions need to be taken prior to starting treatment including polysomnography, checking adrenal gland function, and following IGF1 levels.

What is Prader-Willi Syndrome?

Prader-Willi Syndrome is a disorder of chromosome 15
Prevalence: 1 : 12,000-15,000 (both sexes, all races)
Major characteristics: hypotonia, hypogonadism, hyperphagia, cognitive impairment, challenging behaviors

Major medical concern: morbid obesity

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. Also, 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 a number of the clinical findings should be referred for genetic testing. DNA methylation analysis confirms diagnosis of PWS. FISH and DNA techniques can identify the specific genetic cause and associated recurrence risk. Patients who had negative or inconclusive tests with older techniques should be retested.

- **Recurrence risk** Recurrence is significant only for rare cases with imprinting mutations, translocations, or inversions. All families should receive genetic counseling.

Prader-Willi Syndrome is:

- A non-hereditary birth defect resulting from a disorder of chromosome 15
- A serious, life-long, and life-threatening medical condition
- Occurs in 1:12,000 to 1:15,000 births; both sexes, all races
- Characterized by
  - Hypotonia *(low tone)*
  - Hypogonadism *(underdeveloped sex organs)*
  - Hyperphagia *(uncontrollable hunger)*
  - Cognitive impairment
  - Challenging behaviors
- One of the most common conditions seen in genetic clinics
- The most common genetic cause of life-threatening obesity

A major medical concern is morbid obesity, however with early diagnosis and early intervention, many children can maintain a healthy weight.

Prader-Willi Syndrome Association | USA

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Medical Overview

A Diagnosis and Reference Guide on Prader-Willi Syndrome for Physicians and Other Health Professionals

Prader-Willi
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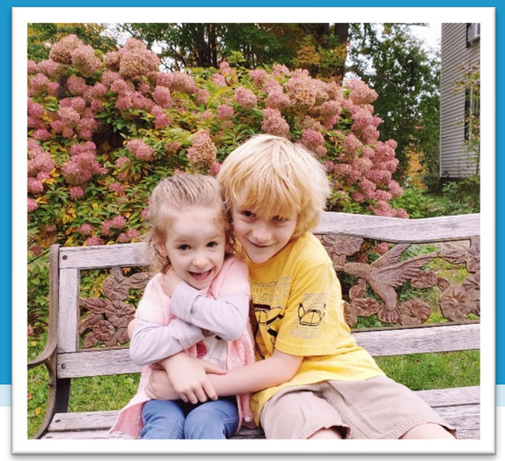

Life Threatening Medical Concerns

- **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 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.
- **Body temperature abnormalities.** Idiopathic hyper- and hypothermia have been reported. Hyperthermia may occur during minor illness and in procedures requiring anesthesia. Fever may be absent despite serious infection.
- **Severe gastric illness.** Abdominal distention or bloating, pain and vomiting may be signs of life-threatening gastric inflammation or necrosis, more common in PWS than in the general population. Rather than localized pain, there may be a general feeling of unwellness. If an individual with PWS has these symptoms, close observation is needed. A CAT scan of the abdomen and/or endoscopy may be necessary to determine degree of the problem and possible need for emergency surgery.
- **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 be supervised at all times in all settings where food is accessible. Those who have normal weight have achieved this because of strict external control of their diet and food intake. 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, stasis ulcers, cellulites, and skin problems in fat folds.

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

Resources for Health Care Providers

"Growth Hormone and Prader-Willi Syndrome, 2nd Edition" and the book "Management of Prader-Willi Syndrome" are available from PWSA | USA at www.pwsausa.org, as are many other publications for professionals and parents. Medical crisis support for professionals and parents is available at (941) 312-0400.

In the Event of Death

Reporting of Deaths
The Prader-Willi Syndrome Association | USA has created a research database of reported deaths of individuals with PWS. In the event of death of someone with PWS, please contact PWSA | USA: (941) 312-0400

Organ Donation for Research
When a child or adult with PWS dies, the family may wish to consider donation of organs for research. Prompt action is essential for tissue preservation. Families are advised to contact the Brain and Tissue Bank directly: 1-800-847-1539 (Maryland).

Our Mission

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

Our Vision

- We will provide **emotional support** to families and caregivers, and nurture hope, strength and connection as we strive together to improve the quality of life of individuals with Prader-Willi syndrome.
- We will take a leadership role in **supporting and stimulating research,** which will ultimately lead to a cure.
- We will provide **education and resources** to **improve the quality of life** for everyone with the syndrome as well as the people who support them.
- We will instill a sense of **compassion and understanding** through our dedication to providing **awareness** and enlightened **knowledge** about the syndrome.
- We will **secure the resources** necessary to accomplish our goals.

Furthermore, **we believe research findings** related to PWS will significantly **impact the understanding** of obesity and appetite regulation in the general population.

Prader-Willi Syndrome is:

- A non-hereditary birth defect resulting from a disorder of chromosome 15
- A serious, life-long, and life-threatening medical condition
- Occurs in 1:12,000 to 1:15,000 births; both sexes, all races
- Characterized by
  - Hypotonia *(low tone)*
  - Hypogonadism *(underdeveloped sex organs)*
  - Hyperphagia *(uncontrollable hunger)*
  - Cognitive impairment
  - Challenging behaviors
- One of the most common conditions seen in genetic clinics
- The most common genetic cause of obesity

A major medical concern is morbid obesity, however with early diagnosis and early intervention, many children can maintain a healthy weight. Prader-Willi Syndrome Association | USA

1032 E Brandon Blvd, #4744
Brandon, FL 33511

Tel: 941.312.0400
E-mail: info@pwsausa.org
Web: www.pwsausa.org

**PWSA | USA, a 501(c)(3) non-profit organization, is very grateful to our donors and invites support of our organization. All contributions are tax deductible to the extent allowed by law.*

We hope you find these materials helpful and that you consider a donation to PWSA | USA to assist in developing more good work(s) like this. Please see our website, www.pwsausa.org.

Helping All Families and Professionals

A Reference to PWSA | USA Support and Services

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

We Offer

Family Support Program

Parent Mentoring

- Parent to Parent support with a trained parent mentor
- Extensive age-appropriate materials provided at no cost
- Free packet for physicians which includes a medical overview of PWS (DVD) and a Growth Hormone booklet.

Crisis Support

- Counseling with trained staff with advanced degrees
- Support for medical, behavioral, legal, school and placement crises
- Information and referral services

Advocacy

- Special education advocacy on behalf of students with PWS including information about IEPs, PWS-specific school strategies, and a DVD for educators.
- Advocacy to help people and families living with PWS connect with needed benefits and services including SSI and essential therapies.
- Support for public policies that increase services and opportunities for people with PWS and their families.

State Chapters

- Chapters offer local advocacy, resources and family connections. To find a chapter in your state, visit: www.pwsausa.org/links/chapter.htm
- A State Leader's Team coordinates partnership with national support services.

Medical and Research Program

- Scientific and Clinical Advisory Boards - all members have extensive PWS experience
- Consultation with PWSA | USA medical board members available to all physicians
- Scientific conference at the annual national PWSA (USA) conference
- Publishing of Medical Alert booklets and medical handouts that are PWS-expert physician authored
- New Third Edition of "Management of PWS" book for professionals
- Research grants awarded annually
- Research advocacy - PWSA | USA was instrumental in acquiring FDA approval for the use of growth hormone for individuals with PWS

Publications and Website

- Extensive website information with downloadable publications
- Bi-monthly educational newsletter for all members, including the latest in medical information, research and treatment
- Educational, supportive, and medical publications including DVDs available
- Moderated PWSA | USA e-mail support groups

Awareness/PR/Development

- Booths/Presentations at medical conferences nationally and internationally
- Annual national PWSA | USA Conference for families and professionals
- Extensive work with national and local media
- "Grass roots" fundraising and chapter efforts
- Annual national PWS Awareness Month
- Support individual awareness efforts
- Awareness merchandise

Bereavement Support and Study of Deaths

- Bereavement support by phone and mailings
- Sponsorship of largest study of PWS deaths

International Support

- Educational materials donated to developing countries
- Reproduction of our extensive education materials in their own languages encouraged
- Parents and professionals worldwide linked to our international organization, International Prader-Willi Syndrome Organization

Opportunity to Contribute

- Volunteers host local fundraisers
- Planned Giving, Corporate Matching and CFC opportunities
- Donations are tax deductible*

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A major medical concern is morbid obesity.

The Prader-Willi Syndrome Association | USA was formed in 1975 to provide a vehicle of communication for parents, professionals, and other interested citizens. It is an organization dedicated to the sharing of experiences in how to cope and work with the syndrome. Chapters of PWSA | USA are located in most states and are available for support, education and advocacy.

PWSA | USA is supported solely by donations. Our agency has made a difference in the lives of many families and individuals affected by this unique syndrome.

YOU TOO CAN HELP. Donations may be sent to the address below.

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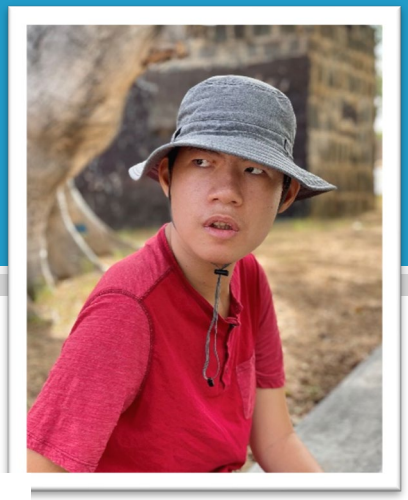
Please see our website, www.pwsausa.org.

Someone You Know Has PWS

An introduction to Prader-Willi syndrome especially for babysitters, neighbors, extended family, friends, play-date occasions, and the community

DID YOU KNOW…



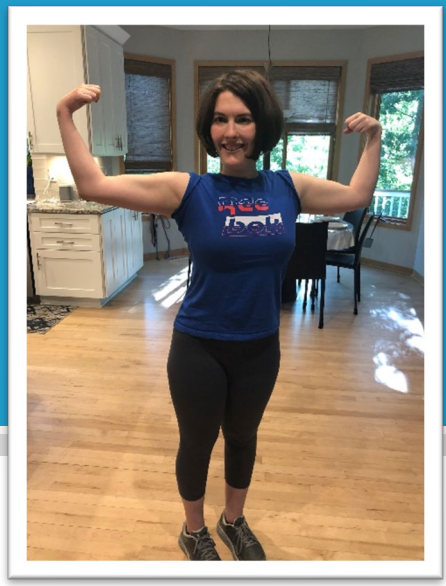
Prader-Willi syndrome (PWS), pronounced PRAH-der-WILL-ee, is a birth defect. This means that a person is born with the syndrome and cannot outgrow it. Doctors don't know why it happens and there is no cure for it.

People who have PWS like to do the same things as other children and adults. They like to go to movies, have parties, play outside, and have hobbies, like putting puzzles together or making bracelets.

Some of the characteristics of children and adults who have PWS are:

- Babies born with PWS are usually weak from lack of muscle tone and have a hard time sucking. It takes a long time for them to learn to walk and to talk clearly. They get stronger as they grow, but have poor balance and are not well coordinated. Toddlers can have a food drive, but this usually occurs after 3 years of age.

- Children and adults have a compulsion to eat that they cannot control. They never feel full and always feel very hungry. The certainty that food is not available frees them up to work and play.
- Children and adults cannot eat as much as everyone else because they will gain LOTS of weight on considerably fewer calories.
- Children and adults who have PWS are usually friendly, pleasant and mild mannered. If they become anxious or distraught, it's very hard for them to settle down. They do not have control over their behavior.
- Some adolescents do not reach full physical maturity. They can be shorter than normal and look younger than they really are.
- Children and adults with PWS are generally concrete thinkers. Terms like, "Hop to it!" may not be understood to mean "Begin the task immediately" and may cause confusion, anxiety, and result in an unwanted behavior.
- People with PWS have a delay in processing the information you give them. Most children will take between 3-5 seconds to understand what you say. If too many instructions are given or the instructions are generalized, they can miss the middle part of what you said and misunderstandings can occur.



YOU CAN HELP BY:

- Never make fun of the child or adult with Prader-Willi syndrome.
- Resist your temptation to give him or her any food, even one cookie or a piece of candy.
- Be aware of their need and ability to tell you made-up stories so that they can have more food.
- Keep food out of sight, out of reach, and never leave the person in a room that has food. The best way is to keep all food locked up. Food can harm and even kill a person who has PWS. Remember, the hand of someone with PWS is quicker than the eye!
- Substitute diet soda, diet gum, or raw fruits and vegetables for foods that have higher calories.
- Understand that a person with PWS cannot resist trying to get food; if they are slim, it is because the family has worked very hard to control their food and weight.
- When someone with PWS becomes upset, allow time and space for them to settle down. Looking at them or trying to talk to them usually only makes them feel worse. Ask their parents or caretaker for tips on what helps them.

Q: How is PWS diagnosed?

A: Suspicion of the diagnosis is first assessed clinically, then confirmed by specialized genetic testing on a blood sample. Formal diagnostic criteria for the clinical recognition of PWS have been published (Holm et al, Pediatrics 91, 398,1993) and further refined (Gunay-Aygun M, Schwartz S, Heeger S, O'Riordan MA, Cassidy SB. The changing purpose of Prader-Willi syndrome clinical diagnostic criteria and proposed revised criteria. Pediatrics, 108(5):E92, 2001). The diagnostic criteria and also laboratory testing guidelines for PWS can be found on the PWSA | USA website www.pwsausa.org, as well as in the 3rd Edition of our "Management of Prader- Willi Syndrome" textbook. Genetic testing is now very accurate for the diagnosis.

Q: What is known about the genetic abnormality?

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

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

A: PWS is a complex genetic disorder that typically causes low muscle tone, short stature, incomplete sexual development, cognitive disabilities, problem behaviors, and a chronic feeling of hunger that, IF FOOD ACCESS IS NOT RIGOROUSLY CONTROLLED, can lead to excessive eating and life-threatening obesity.

Q: Is PWS inherited?

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

Q: How common is PWS?

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

The Prader-Willi Syndrome Association | USA was formed in 1975 in order to provide a vehicle of communication for parents, professionals and other interested citizens. It is an organization dedicated to the sharing of experiences in how to cope and work with the syndrome. Chapters of PWSA | USA are located in most states and are available for support, education and advocacy.

A national conference, a bi-weekly newsletter entitled *The Pulse,* which incorporates contributions from members as well as professional advice, and a wealth of other written materials and publications on Prader-Willi syndrome are all part of the PWSA | USA's effort to carry out its mission.

PWSA | USA is supported solely by membership fees and donations. Our agency has made a difference in the lives of many families and individuals affected by this unique syndrome. **YOU TOO CAN HELP.** Donations and membership applications may be sent to the address below.

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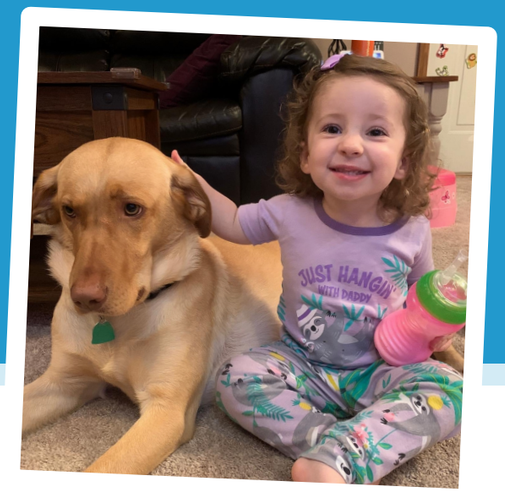
Questions and Answers

About Prader-Willi Syndrome

Prader-Willi SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

Our Mission

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



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

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

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

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

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

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

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

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

Q: Does early diagnosis help?

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

Q: How does growth hormone therapy help?

A: Use of growth hormone therapy is becoming standard care for those with PWS, when prescribed with appropriate precautions by an experienced endocrinologist. Growth hormone therapy can help with height, weight, body mass, strength, and agility, among other areas. It may also help with cognitive development. Reports have indicated dramatic increase in growth rate (especially in the first year of treatment) and a variety of other effects, including improved body composition (higher muscle mass, lower fat mass); improved weight management; increased energy and physical activity; improved strength, agility and endurance; and improved respiratory function.

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

A: With help, people with PWS can expect to accomplish many of the things their "normal" peers do - complete school, participate in their outside areas of interest, be a productive worker under the right conditions, even move away from their family home. They do, however, need a significant amount of support from their families and from school, work and residential service providers, both to achieve these goals and to avoid obesity and the serious health consequences that accompany it. Even those with IQs in the normal range need lifelong diet supervision and protection from food availability. Although in the past many people with PWS died in adolescence or young adulthood, prevention of obesity can offer those with the syndrome the probability of living a normal life span.

New medications, including psychotropic drugs and synthetic growth hormone, are already improving the quality of life for some people with PWS. Ongoing research offers the hope of new discoveries that will enable people affected by this unusual condition to live more independent lives.

PRADER-WILLI SYNDROME
MEDICAL ALERTS

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

941.312.0400 | www.pwsausa.org

REVISED 2022

Prader-Willi
SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

PWSA | USA Mission

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

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

Revised 2022

TABLE OF CONTENTS

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

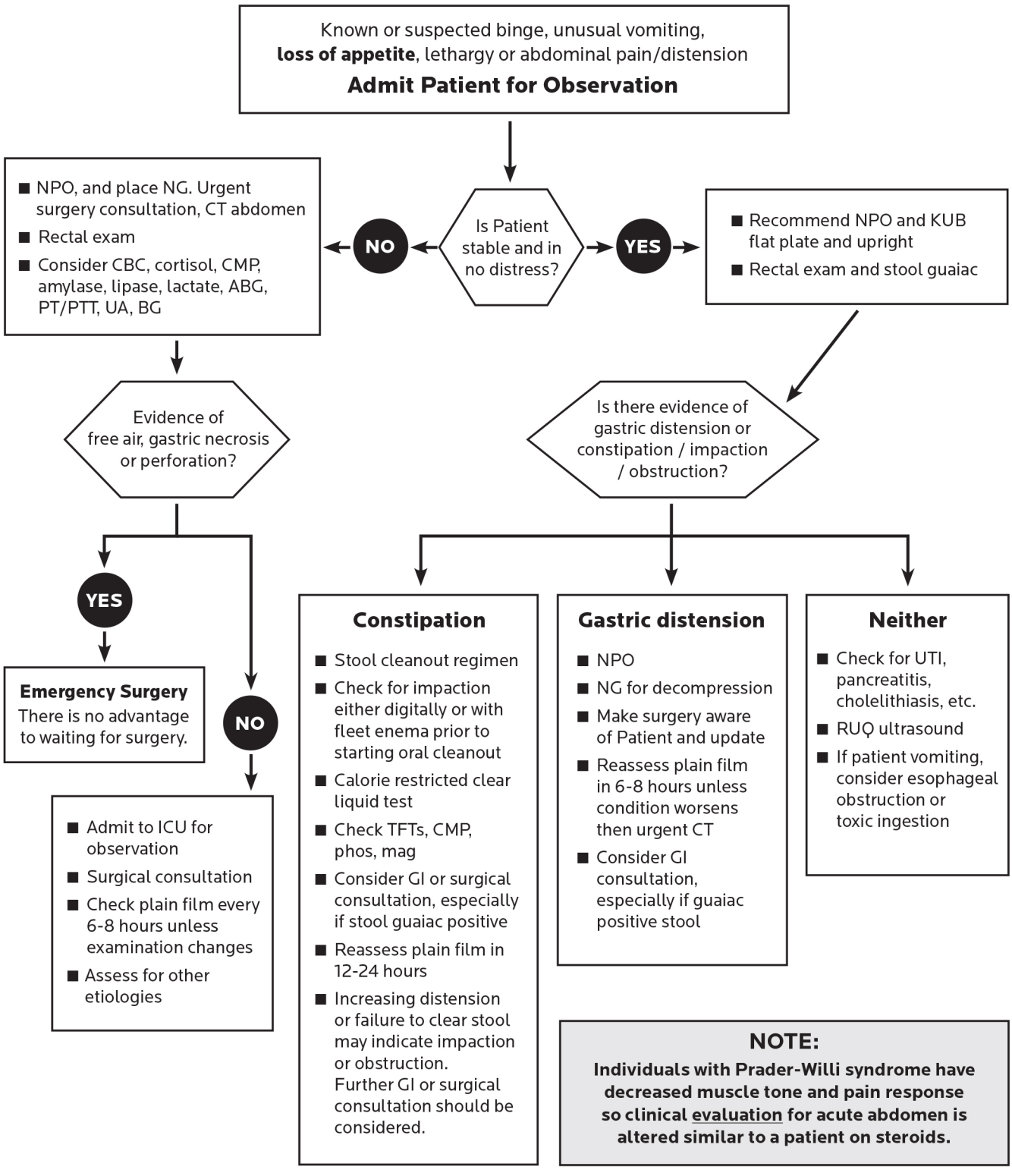
PWSA | USA collects information on mortality to help advance knowledge about causes of death. That knowledge can lead to future research and address critical areas of need for advancing treatment development and quality of life/longevity for those with PWS. We also partner with Autism BrainNet to facilitate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

Evaluation of Individuals with Prader-Willi Syndrome with New GI Complaints



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Prader-Willi SYNDROME ASSOCIATION | USA
SAVING AND TRANSFORMING LIVES

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Clinicians of the PWSA I USA Clinical Advisory Board
and consultant experts

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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