Prader-Willi Syndrome

Basic Information And Medical Concerns

Debbie Mason, RN PWS Care Manager
Objectives

- To provide education about Prader-Willi Syndrome (PWS), a rare and complex genetic disorder
- To provide information on medical alerts regarding patients with PWS
- To provide references and medical contacts that are available for more information on PWS
What is Prader-Willi Syndrome

- A complex genetic disorder affecting appetite, growth, metabolism, cognitive function, and behavior.
- Abnormal chromosome 15 q11-q13
- 3 possible genetic mechanisms – deletion, disomy, or imprinting error
- Autosomal – equal occurrence in both genders
- 1 in 12,000-15,000 births
Features of the Syndrome

- Hypotonia
- Short stature, small hands and feet, and almond shaped eyes
- Hypogonadism
- Hyperphagia
- Low metabolism
- Obsessive compulsive tendencies, often associated with food
- Cognitive delays
- Social and motor deficits
Two Distinct Phases

- FAILURE TO THRIVE: Infants are low birth weight, exhibit weak suck, hypotonia, and have feeding difficulties in the first year of life.

- OVER-THRIVING: Begin to thrive too well, onset of hyperphagia. Usually around age 2.
  - Obesity occurs if not managed
Behavioral Issues

- Food seeking
- Food stealing
- Difficulty with change/transition – a structured routine is very important
- Verbal/physical outbursts and aggression involving food and change to routines
- Very sensitive to emotional environment
- Skin picking
- Obsessive compulsive behaviors
- Mood lability
Medical Alerts in Prader-Willi Syndrome

- Can have a high pain threshold
- Rarely will be able to vomit
- Body temperature abnormalities
- Skin lesions – open sores from skin picking
- Respiratory concerns due to hypotonia (weak chest muscles) and sleep apnea
- Possible adverse reactions to medications and anaesthesia
- Hyperphagia – insatiable appetite
Medical Alerts in an Inpatient Setting

- **Cognitive Limitations** — use simple, understandable language

- **Hyperphagia (insatiable appetite)** — Can go to extreme measures to obtain food (age related). Must be on a calorie restricted diet. Food sources must be monitored and controlled.

- **Adverse Reactions to Anesthesia and Meds**
  Use extreme caution in giving medication that may cause sedation. Prolonged exaggerated responses, including respiratory arrest, have been seen. Be conservative in administering pain medications.
Inpatient Alerts - Continued

- **Respiratory Concerns** — at risk for respiratory difficulties. Sleep apnea is common. Keep on pulse oximetry for 24 hours. If they use CPAP at home, must be put on CPAP when extubated.

- **Altered Pain Response** — Can have a very high pain threshold which may mask injury, infection or illness. Do not use pain as a primary means of diagnosis. Some may not complain of pain until infection or injury becomes severe.

- **Body Temperature Abnormalities** - due to hypothalamic malfunction. Hyperthermia may occur during minor illness or fever may be absent in cases of severe infection.

- **Skin Lesions** — skin picking is a common reaction to stress. Extra measures to cover incisions, IV sites and/or wounds should be used.
A recent article in the Journal of Clinical Endocrinology and Metabolism (de Lind van Wiingaarden RF, Otten BJ, Festen DAM, et al. High Prevalence of central adrenal insufficiency in patients with Prader-Willi syndrome. J Clin Endocrinol Metab. 2008 May;93(5):1649-54) indicates that there may be a high frequency of central adrenal insufficiency in PWS individuals. This only becomes apparent during stress. The recommendation at this time is to treat the patient with stress-dose steroids (hydrocortisone) during times of illness/surgery.

Dr. Jennifer L. Miller, Endocrinology, University of Florida, who follows over 100 patients with PWS, recommends to empirically treat with steroids before surgery and until they are eating normally.

Please check the PWSA (USA) web site for updated information.
Guidelines for Post Operative Monitoring

- Increased morbidity after surgery due to:
  - Abnormal physiologic response to hypercapnia (high levels of CO2) and hypoxia
  - Hypotonia
  - Narrow oropharyngeal space
  - High incidence of central, obstructive and mixed apnea
  - Thick oral secretions
  - Obesity
  - Increased incidence of scoliosis with decreased pulmonary function
  - Prolonged, exaggerated response to sedatives
  - Increased risk for aspiration
Recommendations

- Infants and children with PWS who undergo deep sedation and general anesthesia should undergo recovery in a monitored unit, either the Pediatric Recovery Room or PICU.
- Continuous pulse oximetry must be used for 24 hours post op, with attention to airway and breathing.
- Conservative approach to pain management and use of narcotics.
- Full assessment of GI motility should be performed before allowing patient to eat/drink because of the predisposition to ileus after surgery.
- Direct supervision (1:1) is essential to prevent foraging for food postoperatively.

(recommendations from Winthrop University Hospital, Mineola, New York, Primary Teaching Affiliate of the SUNY at Stony Brook School of Medicine, who have cared for over 300 patients with Prader-Willi Syndrome)
Medical Contacts

- Dr. David Viskochil, University of Utah, Medical Genetics  
  (801) 587-7689
- Dr. David Donaldson, University of Utah, Pediatric Endocrinology  
  (801) 587-3922
- Dr. Kathleen Pfeffer, Sleep and Pulmonary Specialist  
  (801) 967-3155
- Dr. Philip Baese, University of Utah, Neurobehavior HOME  
  Psychiatry (801) 581-8110
  (These physicians are providers at the PWS Multidisciplinary Clinic)
- Debbie Mason, Care Manager/Clinical Care Specialist,  
  utahpwsa@gmail.com, (801)712-0501
- National PWSA 24/7 Emergency Medical Crisis Hotline (800) 926-4797
Sources of Information

- National Prader-Willi Syndrome Association
  www.pwsausa.org
- Utah Prader-Willi Syndrome Association
  www.upwsa.org
- Management of Prader-Willi Syndrome,
  Third Edition, (Butler, Lee, and Whitman, editors)