

PWSA SCHOOL SUCCESS KIT

INDIVIDUALIZED EDUCATION PROGRAMS:

Medical Overview for School Nurse and Medical Support Staff



Medical Overview for School Nurses and Medical Support Staff

Cause and Diagnosis of PWS

- **PWS occurs from three main genetic errors.** Approximately 70% of cases have a non-inherited deletion in the paternally contributed chromosome 15. Approximately 25% have maternal uniparental disomy (UPD) - two maternal chromosome 15s and no paternal chromosome 15. Additionally, 2-5% have an error in the “imprinting” process that renders the paternal contribution nonfunctional; rarely, these imprinting defects may be inherited.
- **Diagnostic testing** Individuals who have several the clinical findings should be referred for genetic testing. DNA methylation analysis confirms diagnosis of PWS. FISH and DNA techniques can identify the specific genetic cause and associated recurrence risk. Patients who had negative or inconclusive tests with older techniques should be retested.

Life Threatening Medical Concerns

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

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

Potential Characteristics

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

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