Prader-Willi Syndrome

Health Watch Table

1. Head, eyes, ears, nose, throat

Considerations	Recommendations
Children: ▸ Strabismus and myopia are common	▸ Arrange an auditory brainstem response (ABR) in newborns ▸ Undertake ophthalmology evaluation before 2 years of age, with particular attention to strabismus and visual acuity
Considerations	**Recommendations**
Adults: ▸ Visual acuity is more commonly diminished than in the general population	▸ Perform office-based screening of vision annually as recommended for average-risk adults, and when symptoms or signs of visual problems are noted, including changes in behavior and adaptive functioning. ▸ Refer for vision assessment to detect glaucoma and cataracts every 5 years after age 45

2. Dental

Considerations	Recommendations
Children: ▸ Decreased and sticky saliva flow can predispose to dental caries ▸ Delays in teeth eruption and dental overcrowding may occur	▸ Attend to oral hygiene in infants and children including use of soft foam toothbrushes, as well as dental products (toothpaste, sugarless gums, mouthwash) to stimulate saliva production ▸ Arrange regular dental visits with particular attention to crowding of teeth and dental caries ▸ Make orthodontic referral, as necessary

3. Cardiovascular

Considerations	Recommendations
Adults: ▸ *Cor pulmonale* is a commonly reported cardio-vascular complication in those who are obese or have significant obstructive sleep apnea (OSA) ▸ Cardiopulmonary compromise related to obesity is a common cause of death ▸ Hypertension is frequently reported but is uncommon in children	▸ When any risk factor is present, screen for cardiovascular disease earlier and more regularly than in the general population and promote prevention (e.g., increasing physical activity, reducing smoking) ▸ Arrange cardiac evaluation, including cardiology consultation, for severely obese patients. ▸ Manage underlying obesity (see below)

4. Respiratory

Considerations	Recommendations
Children: ▸ Unexpected death may be caused by respiratory obstruction early in growth hormone therapy ▸ Upper respiratory tract infections may affect some children and adults significantly.	▸ Refer to ENT for evaluation for removal of tonsils/adenoids, if obstruction is present ▸ All patients with PWS who have an upper respiratory tract infection or other respiratory symptoms should be assessed as soon as possible ▸ Ascertain a sleep history and then arrange a sleep study before anesthesia, and if evidence of respiratory distress, sleep apnea, or obesity is present
Adults: ▸ Cardiopulmonary compromise is the most common cause of death	▸ Ascertain a sleep history with attention to sleep disorders, obesity of any level, snoring, asthma, respiratory infections, and excessive daytime sleepiness ▸ Consider cardiology or pulmonary referral, as needed

5. Sleep

Considerations	Recommendations
Children: ▸ At risk for sleep-disordered breathing	▸ Arrange routine sleep studies during infancy and childhood, and before starting growth hormone therapy and three months after initiating therapy ▸ Ascertain a sleep history and arrange a sleep study before use of anesthesia, and if evidence of respiratory distress, sleep apnea or obesity is present.
Children and Adults: ▸ Narcolepsy/cataplexy is more common than in the general population ▸ At risk for sleep paralysis upon falling asleep or awakening, which may include hallucinations	▸ Evaluate for daytime sleepiness or loss of muscle tone provoked by excitement or other strong emotions ▸ Ascertain a sleep history, with attention to sleep disorders, obesity, snoring, asthma, respiratory infections, and excessive daytime sleepiness
Adults: ▸ Continue to be at risk for sleep-disordered breathing	▸ Consider a sleep study, pulmonology, and ENT referral, as indicated

6. Gastrointestinal & Nutrition

Considerations	Recommendations
Children: ▸ Early concerns include gastroesophageal reflux disease (GERD) and reduced intake due to poor sucking ▸ Failure to thrive is common in infancy followed by the development of hyperphagia and obesity in early childhood ▸ ~10% develop gall bladder stones ▸ Gastroparesis is common	▸ Ascertain a comprehensive GI and nutrition history ▸ Undertake video swallow in neonates based on clinical concerns ▸ Attend to feeding ability and need for assisted feeding ▸ Educate caregivers regarding the necessity of a lower-calorie regime, and environmental controls to prevent ready access to food ▸ Attend to diet, nutrition, physical activity, and obesity, including plotting weight on standard growth charts ▸ Refer to a dietitian/physician with experience in PWS, if possible, to develop an appropriate nutrition and food security regime ▸ Refer to a gastroenterologist, nutritionist, or dietician, as appropriate. Behavioral management programs should be instituted

6. Gastrointestinal & Nutrition (continued)

Considerations	Recommendations
Adults: ▸ Obesity is common without a nutrition and food security program ▸ Vomiting often reflects very serious illness (e.g., gastric necropsy) ▸ Gastroparesis is common ▸ Anal picking is common and may lead to colonic tears/ bleeding ▸ Constipation due to hypotonia is common	▸ Ascertain a comprehensive GI and nutrition history. Attend to diet, nutrition, and obesity. Refer to a gastroenterologist, dietitian/physician with experience in PWS. Implement the modified Red, Yellow, Green (RYG) 2 diet based on energy requirements (ideally measured by indirect calorimetry) and food security programs ▸ Behavioral management should be maintained with the assistance of a behavioral specialist ▸ In the event of emesis history, the adult with PWS requires immediate evaluation and possibly medical imaging ▸ Recommend daily multivitamins ▸ Provide usual interventions to prevent and manage constipation

7. Genitourinary

Considerations	Recommendations
Children: ▸ 80%-90% of males have cryptorchidism ▸ Precocious adrenarche may occur ▸ Delayed and incomplete pubertal development is common in both sexes	▸ Verify testicular descent before 2 years of age ▸ Refer to a urologist for cryptorchidism (i.e., absence of one or both testes from the scrotum) ▸ Consider referral to an endocrinologist or gynecologist/urologist, as appropriate, regarding hormone replacement therapy (HRT)
Adults: ▸ Incomplete pubertal development is common in both sexes	▸ Refer to gynecologist/urologist, as indicated by clinical findings, and for guidance regarding HRT for both sexes

8. Sexual function

Considerations	Recommendations
Adults: ▸ Males and most females are infertile ▸ Pregnancy, though unlikely, has been reported	▸ Educate and, if sexually active, counsel ▸ Consider contraception in women who menstruate

9. Musculoskeletal

Considerations	Recommendations
Children: ▸ 30%-70% have scoliosis ▸ 10% have hip dysplasia ▸ Prevention of osteoporosis should start at an early age	▸ Assess for hip dysplasia in early infancy and before 2 years of age ▸ Evaluate for scoliosis from infancy ▸ Monitor with X-rays and refer to an orthopedic surgeon as necessary (Timing of surgical interventions are influenced by the severity of scoliosis and the degree of skeletal maturation) ▸ Ensure adequate intake of calcium and vitamins D3 and K from childhood ▸ Encourage a weight-bearing exercise program

9. Musculoskeletal (continued)

Considerations	Recommendations
Adults: ▸ Scoliosis and osteopenia/osteoporosis are common in both sexes. ▸ Kyphosis may also occur	▸ Screen for scoliosis and kyphosis with spinal X-rays and refer to an orthopedic surgeon as necessary ▸ Assure adequate calcium and vitamins D3 and K intake ▸ Screen for osteoporosis with regular bone mineral density tests ▸ Refer to an endocrinologist for consideration of sex-hormone therapy to promote bone health

10. Neurology

Considerations	Recommendations
Children: ▸ Hypotonia is common and leads to impaired or absent swallowing and sucking reflexes ▸ Hypotonia gradually improves over time ▸ 10% have epilepsy ▸ All have some degree of cognitive impairment	▸ Undertake clinical evaluation with attention to reduced motor activity and psychomotor delay ▸ Consult relevant specialists as indicated by clinical findings ▸ Treat epilepsy as in general population

11. Behavioral/mental health

Considerations	Recommendations
Children & Adults: ▸ Severe skin picking is common and tends to increase with age ▸ Severe maladaptive behaviors are common (including obsessive-compulsive disorders). ▸ Psychosis may occur in adolescents and adults. Some features of PWS (e.g., tantrums, aggression, compulsivity, anxiety and mood disorder) may be treated with specific pharmacological agents ▸ Risperidone, if indicated, does not usually lead to additional weight gain	▸ Examine skin for evidence of severe skin picking, edema and skin breakdown ▸ A behavior management program is required to support their dietary requirements. Avoid food-related occupational and educational activities. Refer to a psychologist or psychiatrist familiar with PWS when necessary to assist in distinguishing between behavior problems and psychiatric illness

12. Endocrine

Considerations	Recommendations
Children: ▸ Hypothyroidism, diabetes mellitus (Type II), growth hormone (GH) and sex hormone deficiencies may occur ▸ Growth hormone therapy and strict dietary modifications can normalize body habitus ▸ 60% can develop central adrenal insufficiency	▸ Arrange for a PWS pediatric endocrinologist to assess for GH therapy as soon as diagnosis is confirmed. An orthopedic surgery referral may also be indicated before GH treatment is started ▸ Make ENT referral to evaluate upper airway with regards to enlarged tonsils and adenoids prior to starting GH therapy ▸ Screen before and during GH replacement for hypothyroidism, diabetes, and scoliosis. (See No. 4 Respiratory and No. 5 Sleep sections for other recommended assessments prior to GH replacement)

12. Endocrine (continued)	
Considerations	**Recommendations**
	▸ Beginning at age 2, assess obese children for diabetes mellitus (Type II) ▸ Refer to an endocrinologist as appropriate for consideration of sex-hormone replacement therapy (See No. 7 Genitourinary above) ▸ Undertake cortisol evaluation for all children
Adults: ▸ As per children, growth and sex hormone deficiencies continue to be found ▸ Growth hormone therapy in adults can help to prevent obesity and improve strength and endurance	▸ Undertake clinical assessment with attention to thyroid function, diabetes mellitus (Type II), and hypogonadism ▸ Refer to an endocrinologist, as appropriate, including for consideration of GH and sex-hormone therapy

13. Other	
Considerations	**Recommendations**
▸ Molecular causes of PWS differ (e.g., in order of frequency: deletion, uniparental disomy, imprinting errors) each of which effect recurrence risks and possible clinical manifestations	▸ Refer to a genetics clinic for evaluation and counseling, where appropriate

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Expert Clinician Reviewers

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Additional reviewer was Elizabeth Roof, MA, Senior Research Specialist, Prader-Willi and Williams Syndrome Research Projects, Vanderbilt Kennedy Center, Nashville, TN.

Resources

▸ 10 published Prader-Willi syndrome health care guidelines reviewed and compared (For full list of references, see ddprimarycare.surreyplace.ca/wp-content/uploads/2018/03/HWT_Prader-Willi.pdf) Accessed March 2025.
▸ Prader-Willi syndrome websites that may be helpful for families and support persons: Prader-Willi Syndrome Association USA. pwsausa.org. Accessed March 2025.
▸ Ontario Prader Willi Syndrome Association opwsa.com. Accessed March 2025.
▸ Pittsburgh Partnership, Specialists in Prader-Willi Syndrome pittsburghpartnership.com. Accessed March 2025.
▸ Foundation for Prader-Willi Research fpwr.org/about-foundation-prader-willi-research. Accessed March 2025.

References

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2. Balko K. Red yellow green: system for weight management. Toronto: Ontario Prader-Willi Syndrome Association; 2005.
3. de Lind van Wijngaarden RF, Otten BJ, Festen DA, Joosten KF, de Jong FH, Sweep FC, et al. High prevalence of central adrenal insufficiency in patients with Prader-Willi syndrome. *J Clin Endocrinol Metab*. 2008 May;93(5):1649-54.

Other references consulted:

Deal CL. Tony M. Hoybye C. Allen DB. Tauber M. Christiansen JS. 2011 Growth Hormone in Prader-Willi Syndrome Clinical Care Guidelines Workshop Participants. *Journal of Clinical Endocrinology & Metabolism*. 2013 June; 98(6):E1072-87.