IPWSO
International
Prader-Willi Syndrome
Organisation

Adolescents with Prader-Willi syndrome

Medical care: Overview
Medical care: Evaluation

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MEDICAL CARE FOR ADOLESCENTS WITH PRADER-WILLI SYNDROME
An Overview of Medical Problems for Physicians

Prader-Willi syndrome (PWS) is a complex genetic disorder with neurodevelopmental manifestations and many possible medical and psychiatric complications. When suspected by clinical findings, usually identified in infancy, the diagnosis is confirmed by genetic testing. Precise genetic testing to identify the genotype is needed to determine recurrence risk to families and psychiatric prognosis. The earlier the diagnosis is made, the better informed are the parents and the treatment team so that developmentally appropriate interventions can be implemented.

As with any chronic disease, a medical specialty clinic is beneficial for adolescents with PWS to foster an ongoing relationship and communication between the patient/family and medical specialists. The need for supervision and environmental control to manage phenotypic behaviors associated with PWS comes into conflict with the typical strivings for independence during adolescence. Some countries are fortunate to have a PWS clinic where adolescents with PWS are seen by specialists on a yearly basis. Visits with the primary physician should occur every 4-6 months. IPWSO recognizes that access to health services and to specialists may vary considerably due to factors that include distance, transportation, financial resources, or availability. This document summarizes the main health needs for adolescents with PWS. The reader is directed to the other overview and evaluation guidelines in this series for infants with PWS (up to 3 years of age), children with PWS (3-12 years of age), adolescents with PWS (13 years of age and older), and adults with PWS.

Most Common/Significant Medical Findings in Adolescents with PWS:

- Most adolescents with PWS will display all the characteristics of the syndrome including hyperphagia (the uncontrollably strong, biologically-determined drive to seek and eat food), excessive and repetitive behaviors, tantrums and disruptive behavior under stress, cognitive rigidity and difficulty with transitions. Skin picking and other compulsive behaviors can be seen. Food control may be more difficult for some adolescents due to their striving forautonomy.
- Growth hormone can improve body composition, strength, and stamina, but hypotonia and decreased muscle mass persist to some degree and are associated with decreased physical activity and lower calorie needs. Even if weight is normal for height, the fat to lean body mass ratio is high.
- Weight regulation is managed by control of food intake and monitored by regular weighing (at least weekly). Daily exercise is required for calorieexpenditure.
- Very few people with PWS can control their eating behavior. Calorie monitoring and

restriction is essential to prevent obesity and its medical consequences. Access to food must be controlled by constant supervision or by externally by locking doors, food pantries, refrigerators and/or freezers. This includes controlling access to money to buy or valuables to barter for food. Psychological FOOD SECURITY (knowing the daily schedule for meals and snacks, the food and portion size to be served, and the assurance that access to food will be controlled at all other times) will decrease anxiety and food related behaviors at any age.

- A daily plan that establishes a routine of meals, snacks, activities and exercise across the day helps to decrease anticipatory anxiety (manifested by repetitive questioning) and improve transitions.
- Learning problems and intellectual deficiency are common, and most adolescents have persistent difficulties with motor coordination, body awareness in space, speech articulation, working memory, calculation and complex problem solving. Adaptive function as measured by Vineland Adaptive Behavior Scales is rarely commensurate with intellectual IQ. Deficits in social communication (understanding unspoken expression through inference, gestures and facial expressions), social interaction skills with peers, and judgment may persist into adulthood.
- Hormonal insufficiencies such as growth hormone deficiency, hypothyroidism (25% in adolescence), and adrenal insufficiency (rare), if previously identified, will require ongoing treatment. Premature adrenarche is common and should not be confused with precocious puberty, which is rare. Hypogonadism results in pubertal delay, which may contribute to poor psychological adjustment. Both males and females may benefit from a slow titration of gonadal steroid replacement to initiate secondary sexual characteristics during adolescence, while monitoring for exacerbation of mood and behavior. Some adolescents with severe behavior disturbance will not be candidates for sex hormone replacement. Both growth hormone and gonadal steroids improve bone mineral density.
- Gastrointestinal motility problems occur throughout the gut and include chronic constipation, reflux, rumination, choking, and the risk of catastrophic cascade of gastric distention, necrosis and rupture.
- Dental caries are common due to reduced salivation. Gastric reflux can erode tooth enamel, and bruxism can abrade the tooth surface.
- Respiratory abnormalities can include shallow breathing (due to difficulty taking a deep breath related to hypotonia of intercostal muscles and shear weight of the fat pad over the chest in overweight/obesity) and reduced sensitivity to hypercapnia (elevated CO_2 as determined by blood gases).
- Cardiac difficulties are related to autonomic dysfunction and include reduced heart rate variability and decreased elevation of pulse and blood pressure during exercise.
- Spinal deformities, including scoliosis, kyphosis or kyphoscoliosis, are present in about

40%. Yearly clinical examinations of the spine should be performed, with radiographs performed for even the smallest asymmetry.

- Low bone mineral density for age (Z-score) may be present. Persistent limping or vague complaints of extremity pain should be evaluated with a radiograph. Fracture or stress fracture may be present. Heightened pain threshold and decreased ability to identify the anatomical source of discomfort are characteristic of PWS.
- Sleep problems are common and consist of sleep/wake disturbances, such as difficulty sustaining sleep at night and excessive daytime sleepiness, as well as respiratory problems arising during sleep. Sleep studies may reveal sleep apnea (obstructive and/or central). Continuous positive airway pressure (CPAP) may be required, especially for obstructive sleep apnea that is exacerbated by overweight/obesity. Bi-level positive airway pressure (BiPAP) may be required for central sleep apnea. A multiple sleep latency test (MSLT) is required for a diagnosis of narcolepsy; cataplexy (episodic sudden loss of muscle tone while awake) may occur also.
- Severe obesity interferes with developmental progression and can produce medical complications such as type II diabetes, fatty liver, hypertension, lymphedema, respiratory and cardiac insufficiency, sleep apnea, hypercholesterolemia, metabolic syndrome, skin changes and ulcers, and joint abnormalities.
- Enuresis is common at any age in PWS; it may be related to bladder hypotonia, inability to sense bladder fullness, obesity or sleep apnea.
- Skin picking is common but not universal. Most often it occurs in episodes, and the location and pattern of periodicity that develops in childhood continues throughout life. Skin picking is provoked by idleness or stress. When severe, it can lead to scarring, disfigurementand potentially serious infections. Surgical incisions may become a site of excoriation, and picking interferes with healing, potentially causing severe surgical site infections.
- Rectal picking often starts because of constipation; but it is exacerbated by high stress levels. When severe, it can lead to chronic anemia and misdiagnosis as colitis or inflammatory bowel disease. Vaginal picking has also been reported and can be misconstrued as menses.
- Social skills deficits are due to language processing delay, poor speech intelligibility or egocentrism.
- Mood and behavior problems interfere with school, social adjustment and family life.
- Anxiety is pervasive and may lead to depression, especially during the transition to adolescence, where the desire for independence is contrasted with environmental controls and need for assistance in most aspects of daily living skills.
- Increasing episodes of skin picking, temper tantrums and other behavioral abnormalities are often the consequence of increasing stress, which should be

identified andmanaged.

- Skills to increase coping strategies can be taught to adolescents and caregivers, practiced together, and implemented on cue.
- Appropriate behavior should be reinforced with verbal praise and, if needed, nonfood incentives and rewards.

- Psychosis and/or bipolar disorder can arise unexpectedly or with stress. The incidence begins most often in adolescence and increases with age, especially among individuals who have the maternal uniparental disomy genotype. The development of abnormal mental experiences, such as delusions (usually paranoia), hallucinations or the onset of confusion together with mood instability, may be indicative of a psychotic illness requiring psychiatric evaluation and treatment. A failure to eat or loss of ability to perform grooming or dressing activities in a timely manner is associated with psychosis. Catalepsy (an immobilized, trance- like state with lack of response to environmental stimuli) and catatonia (a major psychomotor disorder with alteration of consciousness) may occur; both conditions are associated with waxy flexibility and muscular rigidity, which may be decreased in PWS due to hypotonia.
- Mood and behavioral activation can be induced iatrogenically by medication treatment with SSRIs, some atypical antipsychotics, and/or stimulants. A gradual increase in goal directed behavior or intensification of typical behaviors may indicate a mood shift. Impulsive self- injury, sudden change of thinking or behavior, loss of interest in food or failure to eat may indicate physical or psychiatric illness that requires emergency evaluation.

Additional Issues:

- Adolescents with PWS are unlikely to vomit, and they may not be able to accurately communicate how they feel ill. A change in level of alertness or behavior may be the best indicators that the adolescent is sick. On the other hand, they can present with a variety of somatic complaints, especially if they have learned that going to the doctor is a way to get food, medicine, or attention.
- Central temperature regulation is faulty in PWS due to hypothalamic dysfunction, resulting in either hyperthermia (fever of unknown origin) or a lack of febrile response even with severe infections. Hypothermia has been reported, often exacerbated by some medications, and can lead to death. Because individuals with PWS have deficient *peripheral* body temperature sensation, they may use water for bathing or showering that is too hot with risk of scalding, or they may swim in water that is too cold and turn blue. They may wear too little clothing in the cold weather and too much clothing when it is warm. Also, they may become emotionally attached to specific articles of clothing and resist change. Often education, coaching and supervision is required to teach appropriate clothing selection to match the ambient temperature.

- Pain threshold is high, and they may not be able to locate the site of discomfort. This increases the risk of underappreciating the presenting symptoms and overlooking diseases and fractures. Radiographs (X-ray) must be considered even if symptoms observed are notsevere.
- Gastroparesis and gastric necrosis can occur and is life-threatening. A distended stomach can be seen both after overeating and with other conditions, such as constipation, dietary change or gastroenteritis. Gastric necrosis can occur with a distended stomach, and symptoms may be few. A change in behavior and abdominal distention are early signs. This may progress to breathing problems. Vomiting is a late sign and frequently may indicate life threatening intra- abdominal disease. Medical evaluation may include abdominal radiograph, computerized axial tomography (CT scan), and prompt intervention. Decompressing the stomach with a nasogastric tube can be lifesaving.
- Obesity hypoventilation due to morbid obesity can occur in adolescence. It is associated with dyspnea on minor exertion, obstructive sleep apnea with hypoxemia during sleep, and rising non-pitting edema which culminates in right heart failure. Due to the extent of disability at this stage, adolescents may be wheelchair bound, confined to a lounge chair, or sit up in bed because they are unable to recline due to respiratory compromise. Tracheostomy only complicates and prolongs recovery. Supplemental oxygen greater than 1 liter per minute can suppress ventilatory drive due to decreased sensitivity to hypercapnia (CO_2 levels in blood) in PWS. The treatment of choice is intensive rehabilitation with exercise to mobilize fluid, together with protein sparing, calorie restriction.
- Puberty is delayed in the majority of adolescents with PWS. They may express low self-esteem related to their appearance and the delay in maturation of secondary sexual characteristics. Sex hormone replacement can be considered with close attention to change of mood with estrogen and change of behavior with testosterone. Although many adolescents will be placed on sex hormone replacement therapy, there is evidence that some males and females with PWS will develop a normal puberty in their middle 20's.
- Adolescence is the time for anticipatory guidance about issues of sexuality, risk of sexual exploitation in exchange for food, exposure to sexually transmitted diseases, and gender specific issues of fertility. Males have both central and gonadal infertility, and there have been no reports of paternity, although obtaining serum hormones (testosterone, follicular stimulating hormone, inhibin B) and examination of ejaculate for spermatozoa are the definitive tests. Studies have shown that inhibin B is the predictor for fertility; in males with PWS it decreases with age during the developmental years. Females have the potential to ovulate and may conceive. Serum hormone testing (estrogen, follicular stimulating hormone, inhibin B) and ultrasound of the ovaries is recommended. Inhibin B >20 predicts fertility and this must be tested periodically. If the mother has the deletion genotype, the baby has a 50% chance of having Angelman syndrome. If the mother has the UPD

genotype, she will pass on a normally imprinted maternal chromosome 15 and will have the same chance as a non-PWS woman of having a normal baby. In the few reports of completed pregnancies in PWS, the mothers were unable to breast feed or properly care for their infants.

- Many adolescents with PWS have a desire for romance, but the associated interpersonal conflict can be too stressful for them. Most of the time their romantic attachments are pregenital with kissing, hand holding, and role identification as “boyfriend” or “girlfriend”. Often both genders have fantasies of marriage and express the desire to have a baby. This is best handled with psychological substitution (working with animals, “adoption” through monetary support of disadvantaged children, involvement with nieces and nephews). Sometimes adolescents become fixated on a love object (real or imagined) who does not reciprocate their affection. This may require psychological or psychiatric evaluation and treatment.
- The school curriculum should have realistic choices for future vocational endeavors. Jobs in the food service industry should be avoided. Because the risks associated with food access are ever-present, supervised employment is recommended, even for a person of normal weight. A thorough assessment of vocational skills and neuropsychological assets can help to determine the best job for each individual. Often secondary schools prepare students for independent living, including autonomy with money management and use of public transportation, which may not be appropriate for most young adults with PWS.
- Because adolescents have more freedom with exposure to the community and the internet, they can acquire some habits that become compulsive behaviors, such as cigarette smoking and pornography, which are extremely difficult to unlearn. Anticipatory guidance and preventive strategies are recommended.

Medication:

Many adolescents with PWS will have been receiving growth hormone for many years, and many of the typical facial and body dysmorphisms associated with PWS may have normalized. Growth hormone dose is reduced to adult levels when biparental height has been reached or when the person turns 18 and growth hormone is no longer authorized. Some countries permit the use of low dose growth hormone in adults with PWS. Gonadal steroid replacement may be ongoing to manage risk of osteoporosis. Adolescents with PWS may be receiving a variety of psychotropic medications and over the counter supplements. Due to their small lean body mass, increased fat mass, and increased sensitivity to drugs, a low starting dose is suggested, especially for antihistamines and psychotropic drugs.

Respiratory complications can result from the use of standard doses of benzodiazepines or anesthesia during surgery. Gastroparesis has also occurred following surgery, as the gut may not “wake up” as fast as the brain. Medications such as methylnaltrexone may be helpful

post-surgically in blocking the effects of post-operative opioid medications on gut motility. For the few adolescents diagnosed with adrenal insufficiency, it is important to increase cortisol dose during illness.

General remarks:

This document is designed to address the medical problems typically encountered in adolescents with PWS in an effort to reduce serious complications and improve quality of life. A separate IPWSO document addresses evaluations during regular primary care and subspecialty visits.

PWS is due to absence of genetic information on chromosome 15 due to one of three genetic mechanisms (deletion at chromosome 15q11.2-q13; uniparental disomy 15; an imprinting defect on chromosome 15). It is very strongly recommended that the diagnosis is confirmed through genetic testing. A DNA methylation analysis confirms the diagnosis in >99% of cases, but a further work up should be done by a medical geneticist to determine the molecular class. IPWSO can be of assistance in identifying sources of testing. Some other conditions can overlap in signs and symptoms with PWS.

Please also see medical and other information, most of which is written for a lay audience, on the International Prader-Willi Syndrome Organization (IPWSO) website which includes information about family support organizations in over 100 countries: http://www.ipwso.org.

Sources of detailed information about PWS diagnosis, symptoms, evaluation, and management: Pediatrics: www.pediatrics.org/cgi/doi/10.1542/peds.2010-2820 ; Gene Reviews: https://www.ncbi.nlm.nih.gov/books/NBK1330/ and PWSA Medical Alert Booklet: https://www.pwsausa.org/wp-content/uploads/2015/11/newMAbookfinal.pdf

MEDICAL CARE FOR ADOLESCENTS WITH PRADER-WILLI SYNDROME
Evaluation Guidance for Physicians

Prader-Willi syndrome (PWS) is a complex genetic disorder with many neurodevelopmental and behavioral manifestations that emerge across childhood and adolescence. Potential medical and psychiatric complications are most effectively treated early in their course. This guide has been prepared to advise pediatric or medical physicians how to proactively evaluate and clinically identify early signs of problems in adolescents with PWS. The need for environmental control and supervision to manage the phenotypic behaviors associated with PWS come into conflict with the adolescent's striving for autonomy and independence. Potential complications are detailed in the companion document, Overview of Adolescent Problems for Physicians. The examinations and clinical tests listed below are recommended during the annual primary care or subspecialty visit. Test results and communication with subspecialists can be shared with parents during these annual visits. IPWSO recognizes that access to health services and to specialists may vary considerably, but many countries are developing PWS Multidisciplinary Clinics to facilitate family contact with subspecialists. This document summarizes the main health needs that are recommended in the context of available resources. The reader is directed to the other overview and evaluation guidelines in this series for infants with PWS (up to 3 years of age), children with PWS (3-12 years of age), adolescents with PWS (13 years of age and older), and adults with PWS.

Careful medical history with emphasis on the following:

- Assess school progress with emphasis on learning, behavior and social interaction. Review the individual educational plan and address issues related to PWS (food, behavior, exercise, special education, life skills, and vocational planning).
- Assess the current living situation, including adequacy of supervision and level of parental stress.
- Assess weight management by reviewing the home weight chart and the caregiver's strategy for adjusting intake if weight gain occurs.
- Assess the adequacy of dietary interventions to determine whether consultation with a PWS- knowledgeable dietician is necessary for calorie control, nutritional balance, and vitamin/mineral supplementation to assure adequate macro/micronutrient intake such as calcium, 25-dihydroxyvitamin D3, and iron.
- Inquire about access to food in different environments including home, school, vocational programs, and community settings. Inquire about the adequacy of supervision is each setting.

- Inquire about the nature and severity of food related behaviors. Psychological Food Security (knowing the daily schedule for meals and snacks as well as the food and portion size to be served together with controlled food access) will decrease anxiety and food related behaviors at any age.
- Assess the level of physical activity each day by type and duration, especially sedentary activities such as reading or using the computer and other electronics, which may be excessive and require setting time limits. Determine the nature and duration of physical exercise perday and whether exercise utilizes peer involvement (such as Special Olympics, martial arts or dance lessons) and/or family-centered activities (such as walking or hiking). One to two hours per day of physical activity such as walking, swimming, or cycling is recommended. A physiotherapist should determine the safety of using exercise equipment such as treadmills, exercycles, or ellipticals. A desire for competition can facilitate goal- setting and achievement.
- The need for sensory motor stimulation does not diminish with age. Assess the access to activities promoting sensory motor stimulation and integration such as horseback riding, swimming, dancing, drumming, arts and crafts, etc.
- Monitor the degree of scoliosis and the need for radiographs or consultation with orthopedics.
- Evaluate oral hygiene and dentition for signs of dental caries, enamel erosion or wear due to bruxism. Dental inspection and cleaning should occur 2-3 times peryear.
- For GI review of systems, assess the following:
    - Oral competence with bite, chew and swallow.
    - Rate of eating and occurrence of gorging or choking. Implement Pace and Chase (see Attachments).
    - History of gastroesophageal reflux disease (GERD) and/or rumination. Implement Pace and Chase (see Attachments).
    - History of weight fluctuation due to food acquisition, ingestion of beverages, or constipation.
    - History of abdominal distension (with concern for gastric dilatation), possibly associated with foul smelling burps, complaints of abdominal discomfort, and/or limitation or stoppage of intake.
    - Bowel pattern and consistency using the Bristol Stool Chart – ‘make snakes’ (see Attachments). Consider referral to a gastroenterologist for management of constipation.
    - Evidence of rectal picking (description of feces and/or blood on hands,

blood in the toilet), often associated with constipation, stress or excessive time on the toilet.
    - Toilet hygiene – the motor coordination required for wiping can be a challenge and it worsens with overweight/obesity.

- Sleep/wake: Evaluate sleep/wake history and refer to pulmonary or sleep medicine for snoring, sleep apnea, restless sleep, daytime sleepiness and/or night-time wandering. If the person is already using a continuous positive airway pressure (CPAP) or bi-level positive airway pressure (BiPAP) device, determine if they are compliant and whether settings may need to bechecked.
- Urinary incontinence/enuresis: Inquire about urinary incontinence during the daytime or at night. Sensation of bladder fullness is diminished and transmission of the signal for micturition from bladder to brain may be delayed. This can also occur with constipation or sleep apnea. Due to hypotonia, it is common that the bladder will not empty completely after urination increasing the possibility of daytime accidents or enuresis. Gravity can act as a facilitator if adolescent males stand to urinate.
- Behavior: Assess for occurrence and severity of temper tantrums, disruptive behaviors, cognitive rigidity, collecting, compulsive behavior, repetitive asking, lies and theft, impulsive aggression, and/or threats to harm self/others.
- Skin picking: Evaluate the location, frequency and severity of skin picking or other self- injury. Often skin picking occurs in episodes. The location of picking may inform an intervention plan.
- Movement disorder: Ask about stereotypies, tics, dyskinesias, and unusual body postures, especially if the adolescent is receiving a neuroleptic medication. Cataplexy, catalepsy and catatonia may occur.
- Psychiatric symptoms: Inquire about anxiety, mood disorder, and symptoms of psychosis.
- Current medications and doses: Assess for potential drug interactions, side effects and discontinue medications that are no longer necessary, especially those prescribed as needed.

Relevant body examination, including:

- Height, weight; calculate body mass index (BMI, kg/m^2); plot on appropriate growth curves for adolescents with or without growth hormone (see Links at the end of the document)

- Head circumference (relevant for adolescents receiving growth hormone)
- Abdominal circumference (for those with a history of gastroparesis or those receiving neuroleptic medications)
- Vital signs: Blood pressure and pulse rate (BP/P); sitting and standing
- Quality of interpersonal interaction: Comment on alertness, capacity to engage and sustain eye contact, and ability to establish rapport
- Quality of speech (hypernasality, intelligibility) and communication ability (capacity to express wants and needs)
- Dental - inspect teeth for signs of reflux (erosion of enamel), bruxism, decay
- Heart auscultation - evaluate for signs of cardiac insufficiency
- Lung auscultation for irregular ventilation/atelectasis
- Abdominal exam – evaluate for evidence of constipation, hepatomegaly (fatty liver), tenderness
- Back inspection for scoliosis, kyphosis, rod placement
- Gait and joint assessment for mobility problems
- Lower extremity –
    - Inspection of feet, foot position for pes planus, need for shoe inserts (orthotics) or special shoes
    - Examine for signs of leg edema (pitting or non-pitting), ulcers from picking, or infection
- Integument – examine skin, intertriginous folds, perianal areas, active picks, open sores, ulcers, infections, acanthosis nigricans
- Genitals – assess pubertal status, Tanner staging (rarely progresses beyond stage III)
- Rectum – inspect for fissures, rectal bleeding, evidence of rectal picking

Blood tests (yearly):

- Glycosylated hemoglobin (hemoglobin A1C) and fasting blood glucose (recommend fasting blood test at 8am)
- Blood lipids, cholesterol, liver enzymes
- Thyroid stimulating hormone (TSH) and free thyroxine (T4); free triiodothyronine (T3) if possible to obtain
- 25-hydroxyvitamin D3, calcium, phosphorous
- Hemoglobin and hematocrit; white blood cell count and platelet count
- Sodium (Na+), Potassium (K+), creatinine – Hyponatremia may suggest excessive fluid

intake or a side effect from psychotropic medications and/or mood stabilizing anticonvulsants
- Insulin growth factor (IGF-1) for those treated with growth hormone, confirming dose and compliance
- Males – serum testosterone, luteinizing hormone (LH), follicular stimulating hormone (FSH), and inhibin B, if not receiving testosterone replacement
- Females – serum estradiol, luteinizing hormone (LH), follicular stimulating hormone (FSH), and inhibin B, if not receiving sex hormone replacement. Normal serum inhibin B level (>20) indicates potential fertility.

Recommended Clinical Diagnostic Tests:
- Radiograph (X-ray) of back for scoliosis/kyphosis at initial evaluation; yearly from 10-14 years of age and whenever there are clinical signs of a spinal asymmetry. Ask the radiologist to comment on amount of stool in abdomen.
- Bone age (determined by a radiograph of the left hand) is compared to chronological age to determine how fast the skeleton is growing, usually in association with growth hormone treatment.
- Dual energy X-ray absorptiometry (DEXA) every two years; used to measure bone mineral density (for signs of osteoporosis or osteopenia) and to assess body composition (by determining fat mass).
- Polysomnography to rule out sleep apnea, especially with recent weight gain; multiple sleep latency test (MSLT) for excessive daytime sleepiness to rule out narcolepsy; or re-evaluation by sleep medicine for adjustment of settings on continuous positive airway pressure (CPAP) or bi-level positive airway pressure(BiPAP).

Recommended Clinical Consultations and/or Counselling:
- Bi-annual vision evaluation; referral to ophthalmology for suspected refractive error
- Hearing evaluation, if not done before
- Evaluation by an endocrinologist to discuss sex hormone therapy or therapy for osteoporosis/osteopenia
- Discuss need for gynecological care for girls, family values regarding sex, risk of pregnancy, and sexually transmitted diseases (STD's). See Overview of Adolescent Problems for Physicians for a discussion of puberty, fertility, and pertinent counselling issues.

- Make sure the diagnosis of PWS was confirmed by appropriate genetic testing recommended by a medical geneticist. If not, refer to a medical geneticist for testing and family genetic counseling about recurrence risk.
- Dietician (or equal) every 4-6 months, assuring knowledge of PWS requirements for nutrition and weight management
- Physiotherapy assessment of joints, muscles and recommendations for motor activities and suitable exercise for energy expenditure
- Occupational therapy consultation for developmentally appropriate sensory motor stimulation (sensory diet)
- Consider appropriateness of school placement with respect to curriculum, vocational programming and future supportive employment
- Consider referral to psychology or psychiatry for evaluation and treatment of behavior or mood problems
- Discuss need for establishing legal guardianship
- Discuss future residential care options with the parents
- Discuss future financial status and administrative guidance for obtaining government funding and subsidies

Please Note:

Some other conditions can overlap in signs and symptoms with PWS. It is optimal to assure that the diagnosis is correct through genetic testing. A single genetic test, DNA methylation analysis, can conclusively make the diagnosis in >99% of cases, but a further work up should be done by a medical geneticist. IPWSO can be of assistance in identifying sources of testing.

Sources of detailed information about PWS diagnosis, symptoms, evaluation, and management: Pediatrics: www.pediatrics.org/cgi/doi/10.1542/peds.2010-2820; Gene Reviews: https://www.ncbi.nlm.nih.gov/books/NBK1330/ and PWSA Medical Alert Booklet: https://www.pwsausa.org/wp-content/uploads/2015/11/newMAbookfinal.pdf

Links:

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https://www.pwsausa.org/wp-content/uploads/2017/01/HC-GH-treated-girls-2016.pdf

Attachments:

Bristol stool Chart

Pace and Chase Chart