

Prader-Willi Syndrome Finding Psychiatric Help for Your Child

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Introduction

Psychiatric symptoms can emerge in anyone when the ability to cope with stress has been compromised. Coping mechanisms can be adaptive or maladaptive. Coping strategies can be learned, but they are more likely determined by temperamental characteristics called personality traits. Temperament is both genetically determined and shaped by interpersonal experiences. Individuals with developmental disabilities have intellectual deficiencies, brain differences, and/or physical and sensory handicaps which affect their temperament, their capacity to relate to others in their environment, and their ability to acquire coping strategies. They are not only more likely to experience stress, but they are also less likely to be able to cope with stress adaptively. In these individuals psychiatric symptoms may be the way in which they communicate their distress. Internalizing behaviors (anxiety, depression) and externalizing behaviors (frustration, disruption, aggression) are common psychiatric symptoms which indicate distress.

This summary was prepared to provide guidance to parents and other providers of care in seeking out psychiatric services. Clinical information for the consulting psychiatrist is included.

Psychiatric symptoms may also emerge as a manifestation of an underlying psychiatric disorder. Psychiatric diagnoses are made when a specific cluster of symptoms occurs over a long enough time period to alter an individual's level of functioning in their daily environment. The most common psychiatric disorders are anxiety and depression. Another common set of diagnoses originate in childhood and are classified as disruptive behavior disorders. The most severe psychiatric disorders are indicated by the presence of psychotic symptoms defined by hallucinations, delusions, and disordered thinking. Psychiatric disorders can be familial, genetically determined, associated with specific chromosomal abnormalities, or caused by brain injury. They are also frequently triggered by stress. Certain temperamental traits are also prodromal.

Psychiatric evaluation can:

- delineate symptoms
- identify stressors
- recognize the strengths and weaknesses in coping strategies
- indicate the presence of an underlying psychiatric disorder

Recommendations for intervention may include:

- prescriptions for environmental change
- alterations in interpersonal interaction through behavioral modification
- education to teach new coping strategies
- medication to alter the individual's threshold for response to stress
- medication to treat an underlying psychiatric disorder

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Part I

The Psychiatrist's Role

It is the role of the psychiatrist to obtain a thorough data base including developmental, behavioral, family and medical histories. This may involve interviews of family members, teachers, staff and/or other caretakers, as well as a review of records. The psychiatrist will also interview or observe the patient to obtain a mental status examination. This series of questions and assessments allows the psychiatrist to evaluate the patients attention span, concentration, motor function, speech and language, interactional abilities, mood and affect, thought patterns and problem solving abilities. The mental status examination provides a window through which brain function is indirectly explored. The psychiatrist organizes the data and synthesizes a case formulation delineating the nature of symptoms, their etiology and their significance. A differential diagnosis is made, and further diagnostic testing is recommended. Ultimately, multimodal treatment interventions are suggested. Depending upon the expertise of the psychiatrist, some of the treatment recommendations may be referred to other professionals on the mental health team.

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It is not necessary for the treating psychiatrist to have experience with Prader-Willi Syndrome. Because this is a rare disorder, there are few clinicians who have had the opportunity to see large numbers of patients with PWS.

Rather, parents should seek a credentialed psychiatrist (ABPN certified in either Child and Adolescent Psychiatry or General Psychiatry) who has an interest and experience in caring for persons with developmental disabilities and who is willing to listen, read and learn about the syndrome. Above all, parents should seek a psychiatrist who is expecting to follow the patient over time and who requests office visits and follow up phone calls whenever prescribing a new medication or when changing a medication dose.

Typical follow up may be in 1-3 weeks initially and at least monthly thereafter with an office visit depending on the nature of the symptoms and the type of drug prescribed. Some medications require blood testing when first prescribed, when the dose is changed and at less frequent intervals thereafter, perhaps every 3-6 months.

Parents (or other caretakers) aid the psychiatrist by coming to the appointment prepared with information that includes past life events with approximate dates, medication history and the patient's behavioral, medical as well as family history.

Experience caring for persons with developmental disabilities and a willingness to take time with a complex patient are essential qualities in a psychiatric consultant

Part II

INFORMATION TO SHARE WITH YOUR CHILD'S PSYCHIATRIST:

I Heterogeneity of PWS

The behavioral and psychiatric phenotype of PWS is highly variable. This heterogeneity is thought to be at least in part due to different genotypes and IQ differences.

II The "PWS Personality"

Some temperamental characteristics or personality traits are so commonly present to some degree in PWS that they are considered part of the **behavioral phenotype**. *This phenotype becomes exaggerated with psychiatric illness or other stressor.* While these features may be the "background noise" when considering psychiatric diagnoses in patients with PWS, changes in the severity these symptoms are useful clues to changes in mental status or mood.

There are a large number of features to the Prader-Willi personality; we consider it helpful to categorize them into four clusters: food related behaviors, oppositional defiant behaviors, cognitive rigidity and inflexibility, and behaviors indicating anxiety/insecurity

⇒ All persons with Prader-Willi syndrome qualify for an Axis I Diagnosis of Personality Change Due To A Medical Condition (310.1)

Food related behaviors:

- overeating of typical food
- eating atypical food (frozen, raw, spoiled food or pet food)
- sneaking food in the home
- night time foraging in the home
- arguing or manipulating to get food
- tantruming to get food
- opportunistic food theft or pilfering (shoplifting from a store or stealing food from school or work)
- planned food foraging expeditions in the neighborhood or community
- nonconfrontational, invasive food access (breaking locks on cabinets, refrigerator or freezer)
- confrontational food seeking (threatening or aggressing to access food)

Oppositional defiant behavior:

- noncompliance
- argumentativeness
- tantrums
- manipulation
- lying/confabulation

Cognitive rigidity/inflexibility:

- perseveration, "sticky thinking"
- inability to tolerate uncertainty
- difficulty with transitions or changes,
- restricted interests (jig saw puzzles, word searches)
- collecting or hoarding nonfood items
- poor judgment
- egocentrism/ difficulty taking another's point of view

Anxiety/insecurity:

- somaticization
- excessive seeking of medical attention
- constant need for reassurance
- body image distortion
- moodiness
- abnormal pain threshold
- rituals

Skin picking is a common symptom among individuals with PWS. The topography of the skin picking is usually arms, face and scalp; however, it can include rectal picking, picking of the nasal septum, and excoriation of the vaginal mucosa. The etiology of the skin picking is unclear. It is not a manifestation of OCD, nor does it respond to SSRI's or other serotonergic medications.

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III Psychiatric co-morbidities

The following psychiatric disorders appear to occur with increased frequency in persons with PWS

1. **Sleep disorder**

PWS predisposes to sleep abnormalities including sleep disordered breathing, daytime somnolence and narcolepsy. Obesity-hypoventilation responds to exercise and weight loss. Untreated, obesity-related breathing abnormalities are the primary cause of death. Evaluation of obesity-hypoventilation requires sleep studies.

Daytime somnolence is fairly common across all ages and is present in normal weight as well as obese individuals. Occasionally, adolescents present with the classic narcolepsy triad of daytime sleepiness, sleep paralysis, and cataplexy, including hypnogogic and hypnopompic hallucinations. Treatment of narcolepsy is typical using stimulants, modafanil, and possibly tricyclic antidepressants.

2. **ADD and ADHD**

Inattention, distractibility and impulsivity can emerge during the developmental period. Standard assessment tools may miss these symptoms because of low motor activity and obesity. Nocturnal hypoxia due to sleep related hypoventilation with or without obstructive sleep apnea must be considered in the differential when ADD symptoms present in the obese child or adolescent. The treatment package is typical with behavioral, educational, and psychostimulant interventions.

3. **Oppositional defiant disorder and conduct disorder**

Although argumentativeness, noncompliance and tantrums are part of the PWS personality profile, the intensity may warrant a separate diagnosis during the developmental period. Occasionally symptoms of theft, predatory aggression, property destruction, running away, fire setting and cruelty to animals indicate the presence of a conduct disorder. Behavioral and ecoenvironmental interventions are essential, and placement in a PWS facility may be required. Psychotropic medications may decrease underlying symptoms of impulsivity and anger.

4. **Obsessive compulsive disorder**

Although cognitive rigidity, need for structure and routine, hoarding, and rituals commonly occur in PWS, true OCD symptoms of ordering, counting and hygiene compulsions are far less frequent .. OCD symptoms typically respond to SSRI's but there is a high incidence of mood disorders in PWS and the possibility of mood activation must be kept in mind.

5. **Anxiety disorders**

Generalized anxiety, somatoform disorder, and social anxiety can occur and respond to typical psychological behavioral and psychopharmacological interventions.

6. **Mood Disorders**

There is an increased incidence of mood disorders in persons with Prader-Willi Syndrome. The onset may be as early as the 1st decade. Symptoms have ranged from frank bipolar illness with psychotic

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depression or mania to subtler manifestations of increased irritability and mood lability. Some patients demonstrate rapid cycling.

- **Dysthymia and depression**

Dysthymia and depression occur in response to life stress such as separation events across the developmental years. Low self esteem may occur as the individual reaches adolescence; short stature, obesity, delayed puberty, social skills deficits, and continual conflicts over food and exercise create stress and conflict with little mastery. Separation/individuation issues are again challenged as the individual with PWS enters adulthood when job/workshop opportunities and degrees of freedom in the community are limited due to the constant need for structure, supervision and control over food access. Antidepressant medications can be very helpful, but the dose must be titrated slowly and well monitored due to the risk of mood activation.

- **Bipolar mood disorder**

The incidence of all types of this disorder appears to be higher than expected and may indicate a predisposition among persons with PWS. As previously noted depression is fairly common, and mood activation with antidepressants may be the first indication of a cycling disorder. Bipolar I and II have been identified across the developmental spectrum. Younger children may present with intense disruptive behavior and mood irritability. Juvenile mania should also be suspected if a therapeutic response to behavioral interventions has been poor or if mood activation occurs with stimulants. Psychotic symptoms are frequent and must be evaluated with the developmental age of the individual in mind. Mood stabilizers can be very helpful at standard doses. Atypical neuroleptics may augment therapeutic efficacy especially with psychotic symptoms.

7. **Psychosis**

There may be an increased incidence of psychosis in PWS, however most cases appear to be a manifestation of an underlying cyclic mood disorder. Onset may be in childhood and is usually recognized in the context of a major behavioral change. Some patients may not be able to articulate delusional thinking nor to report hallucinations. Stress appears to play a role in the etiology of these symptoms and some patients have been psychotic during a grief reaction or other stressor and subsequently stabilized.

The incidence of this schizophrenia does not appear to be increased in PWS.

8. **Developmental learning disorders**

The majority of individuals with PWS are clustered in the mild range of mental retardation. A wide variability in learning styles, language skills, visual perceptual skills and nonverbal problem solving skills have been noted. Articulation disorder and dysfluencies are common in children and often persist into adulthood. Because puzzle solving abilities tend to be well developed among many children with PWS non verbal learning disabilities (NVLD) are remarkable when present. Additional symptoms associated with NVLD are pragmatic language deficits, dysgraphia, math disability, and processing problems. Exceptional difficulties "reading" social situations even in otherwise high functioning patients may be a source of behavioral problems.

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9. **Impulse control disorder**

Serious assaultive and destructive acts in PWS are the exception but commonly occur in concert with other psychiatric diagnoses. The degree of aggression is out of proportion to the precipitating event and can be followed by the appearance of fatigue.

This is the appropriate diagnosis for patients in whom the picking behavior is problematic or severe. As previously noted the skin picking associated with PWS is not an OCD symptom and does not respond to conventional medication management.

10. **Factitious disorder**

Patients with PWS typically lie to get out of trouble, often lie to manipulate and some are creative confabulators. They are capable of confabulation of abuse, claims of romantic entanglements, and calling 911 with false reports. They often *like* being in the hospital because of food access and attention. Some individuals fabricate circumstances to obtain an admission. Despite their general noncompliance, they derive satisfaction from being the subject of medical procedures. Similarly they have medication seeking behaviors that are not related to drug abuse. They can be very resistant to changes in their medication regimen. They may complain of pain to avoid work or exercise but their pain threshold appears to be abnormally high. Any review of systems for medical or psychiatric differential must be conducted in an indirect fashion and with corroboration by outside sources to maximize accuracy and to minimize false positives on exam. Massive denial about personal limitations and responsibility for one's actions is nearly universal and sometimes borders on the delusional.

III **Management**

- The choice of psychotropic medication is much the same as in non PWS patients and is *driven by psychiatric diagnosis not by PWS diagnosis*. As in the general population, some patients respond well to one medication and others do not .
- Appetite stimulation from medication is a potential problem for some patients living in a setting where food is not completely controlled. However the neuroleptics, atypical neuroleptics and valproic acid have all been used effectively without a discernible change in food-seeking behavior.
- Attempts to suppress appetite with medication has never had long term success, as far as we know.
- Skin picking is not a manifestation of OCD and does not respond to SSRI's. Topiramate and naltrexone have been reported to be useful but the proportion of responders is not at all clear. Even when picking is severe the behavior can resolve spontaneously or it may last for years with or without serious medical complications. Behavioral programs can be helpful but often "backfire" by if they draw increased attention to the behavior. Rewards for "not picking" are generally

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ineffective and difficult to implement. Patients may respond to a reward for healing of the lesion and they appear to be aided by frequent application of a greasy antibiotic ointment which reduces the ability to pick effectively. Fingernails should be kept short. We have rewarded patients for cooperating with wearing protective bandages or nighttime mitten restraints. One patient achieved rapid healing by wearing heavy pantyhose protecting his lower leg lesions. Other patients have torn through plaster casts in order to pick. Supervision and environmental controls are essential to reduce the amount of tissue damage even though the behavior is likely to continue.

- We do not consider the rectal picking/digging symptom to be a hypersexual behavior; it seems to be a non specific stress symptom. We have definitely seen it improve with treatment of other psychiatric symptoms or with decreased interpersonal conflict. We have not found a sure remedy, either pharmacologic or behavioral. Unnecessary medical testing for rectal or vaginal blood loss can result when this behavior is not recognized.
- Carbamazepine and oxcarbazepine must be used with care. Patients with PWS will sometimes over ingest large amounts of flavored beverages (including non calorie beverages) or even water and appear to be exceptionally prone to hyponatremia.

SSRIs have induced mood activation in a

- SSRIs have induced mood activation in a number of patients causing worsening of behavioral dyscontrol and even psychosis in vulnerable patients. These adverse effects may appear after an initial good response to the SSRI causing physicians to *increase* the dose rather than recognize a treatment emergent effect. Other patients have had their depression or true OCD effectively treated by this class of medication without complication.

Recommended Reading:

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