Knowing Your Patient
A Biopsychosocial Perspective
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Introduction

Throughout this book, a consistent pattern that readers will notice is the inclusion of case vignettes that exemplify the characteristics being examined in that particular chapter. Narrative examples are extraordinarily powerful tools for bringing home a complex issue or a concept that is otherwise difficult to comprehend.

In this chapter, through a long-form case (a “life story” followed by a biopsychosocial formulation), I hope to demonstrate the value of drawing a complete mental image of a patient. It should evoke a holistic, integrated assessment of an individual with Prader-Willi syndrome (PWS) to have a comprehensive approach to the management of a myriad of difficulties a patient might be suffering from.

To get a full understanding of the unique issues faced by the patient and by the caregivers, I recommend obtaining a thorough history that attempts to establish a timeline, a lifelong trajectory with a sense of the major events. This establishes a temporal flow of events, which could be biomedical, psychological, or sociocultural, and which also distinguishes chronic, underlying issues from acute, circumscribed events that might have affected the individual. An evaluation can then focus on the current issues and pathology that are most impairing to the patient and the family, and that hence need to be addressed most urgently. Finally, arriving at an integrated biopsychosocial formulation is the amalgamation of all the information you have gathered, along with your impression on diagnosis, as well as a holistically informed plan of care management.

When I see my patients for the first time, after a brief introduction and setting the frame of the interview, I begin the assessment with a statement: “Give me a sense of time with all its ups and downs – start right from the beginning, and assume I don’t know anything about your loved one or PWS.” This open-ended invitation to tell a story about the patient is not only informative but also establishes a sense of perspective that can allow the care team to have patience with treatment and to prioritize the most important issues, knowing that not all things will be solved immediately.

To exemplify this biopsychosocial holistic approach to the evaluation of a patient with PWS, I present the following case, which is inspired by many evaluations that I have had the opportunity to conduct over the years. Please note that those readers unfamiliar with some of the technical terms and medication details mentioned in the case will learn more about them through the rest of this book. The Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5) is the current standard of classification of mental illnesses. As useful as it is, the behaviors seen in PWS don’t always fit neatly into the
diagnostic criteria described in the DSM-5. As this is an early chapter, rather than focusing on diagnosis and treatment plan, let’s take in the fullness of this individual’s life experience.

### Case Study

Amon is a 24-year-old man with a diagnosis of PWS brought in for a psychiatric assessment by his parents, who were concerned about a recent worsening of sleep issues and anger problems. The patient and his parents were seen together as well as separately during this evaluation. Amon’s parents provided legal guardianship paperwork before their scheduled appointment and he also gave verbal assent to me for this assessment.

In describing Amon’s developmental history, his parents stated that he was born two weeks prematurely. His was “an easy pregnancy” as described by his mother. He was born vaginally and had a weak cry at birth. He was taken home but was noted to be “floppy” and had difficulty sustaining feeding due to an inability to stay on the breast for long. Even when formula feeding was attempted, the bottle would fall off or the suck response was weak. When the parents noticed a delay in weight gain, Amon was taken to a pediatrician who immediately suggested hospitalization for failure to thrive. Amon was in the hospital for two weeks, where he had to endure a nasal feeding tube to sustain nutrition. At this academic institution, given the presentation, a genetic disorder was suspected and a genetic test was conducted. However, the results were not obtained during the hospitalization. A few weeks after discharge from the hospital, the parents met with a geneticist who explained that Amon’s genetic test revealed a diagnosis of PWS. She elaborated that it occurs due to the loss of function of a small part of the long arm of the 15th paternal chromosome through a phenomenon called uniparental disomy (UPD). Amon’s parents were told that this is rare and no one’s fault. It is generally not heritable and occurs completely by chance. The prognosis and all the treatment modalities available at the time, as well as the next steps, were explained. The revelation that Amon had a genetic illness was scary at first, but the initial interaction with the physician who took blame away from his parents and gave them hope was an essential first step to accepting the diagnosis and preparing for the next steps.

A trusting relationship with Amon’s geneticist and pediatrician led to early intervention services. He received physical therapy for low muscle tone and to improve his gait and occupational therapy to improve his dexterity and other fine motor skills. As he got older, he also received intensive speech therapy due to significant speech delays that he has since largely overcome (to the extent that, at the time of this evaluation, other than his hypernasal intonation, Amon did not seem to have any expressive language disability).

Amon’s excellent response to early intervention led to his enrollment in a regular elementary school. In the school setting, he was described as friendly but easily distracted and hyperactive. He was unable to complete tasks without frequent redirection. He failed the second grade and, because of his significant need for redirection, he was kept in the regular education setting but was provided with a 1:1 aide in the classroom.

Amon’s parents worked closely with a nutritionist with expertise in PWS. This led to his parents being very mindful of his diet and caloric intake. Food-seeking behavior was largely absent until age 10. Even after that, his parents quickly instituted rules and limits such as locking the pantry and refrigerator, which were effective in controlling hyperphagia at home. At school, the aide was made aware and his meals were supervised.

Amon had a relatively good experience in elementary school. However, he reports having a “terrible teacher” in the sixth grade. He explains that his teacher then was a “disciplinary.” This was Amon’s first experience of feeling “different” from his peers. This teacher would make no accommodations for his learning delays, in particular his difficulties with the mathematics curriculum. Amon describes feeling bullied and ridiculed by the teacher in front of his peers. This led to further isolation and verbal bullying by his peers for all of the sixth grade. Amon
describes suffering from depression during the sixth and seventh grades. At that time he would often state to his parents that he wished to die. In addition, he exhibited more tantrums, aggression, and had difficulty falling asleep. He began avoiding school and would have frequent outbursts on school days.

When he was 14 years old, Amon had to be hospitalized in an inpatient psychiatric unit as a result of a bout of severe aggression during which he had hit his mother and had tried to run away from home. He recalls that the episode was in the context of him finding out that he would have to go to summer school due to his absenteeism. The thought of being forced to go to school even over the summer break was simply too upsetting to him.

This was Amon’s first experience being away from home and his close-knit family. Amon has vivid memories of his inpatient stay. In particular, he recalls one instance of agitation in the context of wanting an extra serving of food that led to security officers holding him down. “They dragged me to a room and gave me an injection in my behind, and then they locked me up in there alone and left me crying.” The parents further explained that this happened on the first night of his stay in the hospital and his agitation was severe enough that they had no option but to allow the injection to be given in their absence. Despite the explanation, Amon holds a strong negative emotion toward that hospitalization and toward medical settings in general. When asked, Amon reports that he continues to have almost daily nightmares and recollections of bullying from his teacher at school and of his time at the hospital. His parents confirm that he still avoids needles and “needs a sedative” for blood draws. He continues to have a significant startle response and is hypervigilant throughout the day.

Despite Amon’s struggles and genetic illness, his full-scale IQ has been calculated to be 85, which places him in the range of normal intellectual functioning. He is described as high functioning with good reasoning skills. During the evaluation, he comes across as witty and can provide a coherent, albeit circumstantial, account of his past experiences.

Amon’s psychiatric treatment has been sporadic. Before his hospitalization, Amon’s parents were hesitant to bring him to a psychiatrist. In their words: “It’s bad enough that we couldn’t protect him from PWS. We were doting parents who gave him all we could. It felt like a failure to take him to a brain doctor.” They report that there were many signs that he needed help; however, his first encounter with a psychiatrist or therapist (other than the school psychologist) was not until his hospitalization at age 14. Since his hospitalization, he has been under the regular care of both a psychologist and a psychiatrist.

His psychiatric medication history suggests a sensitivity to medications that increase the activity of serotonin. In the past he has been tried on many medications; some mentioned by his parents were fluoxetine, haloperidol, olanzapine, and risperidone. Although he did well with a combination of fluoxetine and haloperidol that was started during his hospitalization, his haloperidol was discontinued shortly after discharge due to side effects. His parents described that “his eyes kept rolling up” with the haloperidol (a phenomenon called oculogyric crisis). However, within two weeks of discontinuation of the haloperidol, Amon suffered from sudden onset of severe aggression, insomnia, irritability, and paranoid thoughts while on fluoxetine alone. Amon started expressing thoughts that were unusual and not based on reality. He would refuse to sleep and paced all night. He would talk in run-on sentences for hours. His parents were especially concerned when Amon started saying “bizarre things.” He was convinced that he was being followed by his dentist from many years ago and that he needed to “get rid of the agents that follow him on the streets.” He would get aggressive if anyone doubted him or tried to confront his thoughts as unreal. These symptoms rapidly resolved with the discontinuation of the fluoxetine and introduction of olanzapine.

Unfortunately, Amon gained 50 pounds within six months of starting the olanzapine. The parents still consider that weight gain to be the tipping point after which Amon’s hyperphagia and weight became hard to control. Although his weight started stabilizing after stopping the
olanzapine and switching over to a new medicine – risperidone – he continues to be obese. His only psychiatric medication at the time of this evaluation was risperidone 2 mg at bedtime.

In addition to medications, Amon continued to receive cognitive behavioral therapy (CBT) once a week. His parents describe Amon as “insightful” and note that he can sometimes use CBT techniques he has learned during therapy to calm himself down. Despite his significant difficulties, once Amon’s teacher changed and he was provided with additional special education services in addition to the fact that he was now in treatment, he started enjoying school again. The structure and routine provided by the school and his after-school engagements helped maintain his mood for several years. His school district recognized his special needs and he stayed in school until the age of 21.

Throughout his childhood and adolescence, Amon remained very attached to his family. Amon’s parents immigrated to the United States from Eastern Europe when they were in their 20s, hoping for a better life for themselves and their future children. Amon’s father had slowly worked his way up and currently works as an accountant. Amon’s mother works at a local grocery store and takes pride in being an involved mother to three children including Amon, who is the youngest. They are Catholic and have a strong connection to the church and their community. Until the age of 22 years old, a year after graduating from school, Amon continued to live with his parents.

Amon’s parents describe that since he “aged out” of the school system at the age of 21, he became increasingly irritable and aggressive at home. Although his parents were initially hesitant to have him live away from them, Amon was now a large adult who was physically hurting them. Through peer-support groups for parents of patients with PWS, they realized the dangers of caregiver burden and started weighing their longer-term options. Eventually, his parents obtained legal guardianship and then were able to locate a residential placement for him. Since being away from home, Amon’s parents describe that he has been moved around from one group home to another at least twice in the past two years. He currently lives in a group home with three other individuals with developmental disabilities. He remains very close to his family. He continues to prefer consistency in caregivers and routines. Despite the time it took to find a stable group home setting for Amon, his parents describe a sense of relief when it took to find a stable group home setting for Amon, his parents describe a sense of relief that he is taken care of for the future and no longer feel guilty about him living away from home.

In discussing Amon’s latest difficulties, his parents report that since his most recent group home placement he is settling in better. However, occasional aggressive outbursts against peers, and rarely against staff, continue to occur. Over the past two weeks, he has also been habitually putting things (such as a piece of paper or plastic) into his nose and ears. This usually occurs when limits are placed upon him such as being asked to stop watching TV. This has led to frequent visits to the local urgent care clinic and even to the emergency room. Amon states that he does not engage in the behavior to harm himself, but he says that he likes to go to the emergency room. He explains, “I like the nurses there and they let me eat whatever I want.”

In addition to the previously mentioned nightmares, he reports that whenever a staff member comes to hold him for any reason, he feels very uncomfortable and sometimes feels like he is back in the “psych unit.” He denies any current suicidal/homicidal ideations and any auditory or visual hallucinations. He appears restless and distracted during the session. He is friendly and able to answer most questions reasonably well, especially when spoken to without his parents. However, he is particularly guarded and becomes fidgety when the topic of his past comes up. He reports missing his parents and siblings when he is at the group home but also says, “I like my freedom and my friends.”

In describing the medical history, his parents report that in addition to having the UPD subtype of PWS, Amon has recently been diagnosed with insulin resistance and has been...
prescribed metformin to help with its management. Shortly into toddlerhood, Amon was started on growth hormone treatment, which he continues at a low dose to this day. He is on these medications in addition to the 2 mg risperidone daily.

On inquiry about family mental health history, the father reports having a sister who has bipolar disorder. There have been no suicides in the family.

Putting Things Together

Amon’s initial presentation is complex and, just like any other patient with PWS, the “whole” of his story is greater than the sum of its parts. A biopsychosocial perspective of his presentation is necessary to ensure a thorough understanding, as well as nuanced management, of his condition. To look at this case in an integrated manner, Figure 1.1 provides an example of a timeline created to highlight the events leading up to his current presentation.

An underlying genetic abnormality affecting the long arm of chromosome 15 via the phenomenon of UPD provides the most significant biological predisposition via a diagnosis of PWS. In particular, patients with UPD are more likely to have higher rates of behavioral problems in comparison to the deletion or imprinting subtypes.\[^3\] Importantly, it is notable that this same abnormality – that is, UPD – provides a better prognosis when it comes to language development and intellect.\[^4,5\] This relative protection from severe intellectual dysfunction, and in particular Amon’s preserved language development, is a biological protective factor and tends to preserve a pattern of higher academic achievement. As is made clear by the history, his diagnosis of PWS is affecting his prognosis beyond the obvious weight gain and metabolic symptoms usually associated with this illness.

The precipitating factor leading to his current presentation is most certainly the continued aggression. This has become an urgent need in the context of increasing difficulty in the management of his symptoms at the various group homes. Importantly, the psychosocial events leading up to his current worsening are likely due to his difficult transition from his parents’ home to a group home. This is compounded by the fact that he has not been in the same home environment for a long enough time to have established a sense of security and predictability.

A biological pattern to look out for is the aspect of increased sensitivity to serotonergic agents as evidenced by Amon’s psychiatric decompensation while on a serotonin reuptake inhibitor (SRI) namely fluoxetine.\[^6\] This limits the ability to utilize this class of medication. In addition, given his diagnosis of PWS, the use of anything that can cause further weight gain is also problematic.

Psychosocially, a perpetuating factor is the current health system structure in the United States, which tapers off quite abruptly the educational and social resources provided to patients who have behavioral or intellectual disabilities. In particular, his “aging out” of special education services and other social services is an important precipitant of his deterioration due to the sudden lack of structure and stimulation.\[^7\]

As you can see from Amon’s case, our patients are unique not only in their clinical presentation but also in their cultural and psychosocial context. The foregoing description is
“FLOPPY BABY”
FAILURE TO THRIVE
PT/OT/SPEECH THERAPY INITIATED
BULLIED IN SCHOOL
OUTPATIENT PSYCHIATRIC TREATMENT
REENGAGES IN SCHOOL – ODD’S WELL
MOVES INTO GROUP HOME
“FLOPPY BABY” MOVED TO GROUP HOME
“FLOPPY BABY” PULLS OUT OF SCHOOL
“FLOPPY BABY” THERAPY INITIATED
PSYCHIATRIC TREATMENT STARTS

كلمة معنوية

AGE
0
3
6
11-13
14-15
16-21
18-24
NORMAL PREGNANCY
0-3
4-6
7-10
11-13
14-15
NORMAL VAGINAL BIRTH
NICU STAY
FIRST PSYCHIATRIC HOSPITALIZATION
GOOD RESPONSE TO EARLY INTERVENTION
HAS DYSTONIA WITH HALOPERIDOL
HAS A DEPRESSIVE EPISODE
OLANZAPINE STARTED FOR PSYCHOSIS
OLANZAPINE STOPPED DUE TO WT. GAIN
SEVERE AGGRESSIVE BEHAVIOR AT HOME
OLANZAPINE STOPPED DUE TO WT. GAIN
FLUOXETINE
OCtid PSYCHOSIS
ENROLLS IN SCHOOL – DOES WELL
MOBILIZES IN GROUP HOME
PHYSICALLY AGGRESSIVE AND TRIES TO ELOPE
ADHD NOTICED IN SCHOOL
PWS (UPD) DIAGNOSED
NICU: neonatal intensive care unit; UPD: uniparental disomy; ED.: education; IEP.: individualized education plan; WT.: weight; NSSI: non-suicidal self injury; ER: emergency room

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simply an example of what a thorough initial evaluation might capture. In this book, we will further examine some of the clinical features described in this case, as well as many others commonly seen in PWS.

Bibliography

Chapter 2
Caregiver Burden in Prader-Willi Syndrome
Carole Filangieri and Deepan Singh

Introduction
Providing care for our loved ones, nurturing them, ensuring their safety, and meeting their needs can be some of the most gratifying and rewarding activities that we do as humans. In general terms, caregivers provide for the well-being of those around them. Taking on the role of caregiver relieves the burden on other family members, allowing them to continue their education or careers. The role of caregiver has traditionally been taken on by women, who are often socialized to caregiving roles as children. As an example, even as children, girls are offered toys that encourage caregiving as pretend play. That said, society often underestimates the role of male caregivers. Approximately 40% of primary caregivers are men. Caregivers tend to children and take care of parents, grandparents, partners, or other family members, friends, and neighbors. Caregiving includes assisting with hygiene and dressing, cooking and cleaning, and providing meals and transportation, as well as scheduling appointments, managing medications, making treatment decisions, and providing emotional support. Caregiving can also provide benefits to the caregiver, including a sense of personal satisfaction—the feeling of being useful to and needed by someone—and it can make life feel more meaningful overall.

Most caregiving is informal. It is unpaid and usually provided by a friend or family member. The Centers for Disease Control (CDC) estimated that the value of unpaid caregiving was approximately $450 million in 2009. In 2018, the value of informal caregivers providing care to loved ones with dementia alone was estimated to be $41 billion. As the cost of healthcare continues to rise, more people are finding themselves shouldering the responsibility of caring for a sick or disabled family member. According to the National Alliance for Caregiving (NAC) and the American Association for Retired People (AARP), in 2020, approximately 53 million American adults identified as unpaid caregivers, with 14.1 million caring for children 17 and under and 47.9 million caring for adults over the age of 18. Notably, 23% of caregivers report that caregiving has caused a decline in their health. Sixty-one percent reported that in addition to providing care to a loved one, they were also employed outside of the home.

Caregiver Burden
As rewarding as caregiving can be, caring for someone with a chronic disorder or disability brings unique challenges that can sometimes leave the caregiver feeling unappreciated and isolated—an experience often characterized as “caregiver burden.” Caregiver burden has been described as “the extent to which caregivers perceive that caregiving has had an adverse effect on their emotional, social, financial, physical, and spiritual functioning,” underscoring the multidimensionality of caregiver burden.
This is particularly true when providing care for someone with Prader-Willi syndrome (PWS). The rarity of the disorder means that many people have never heard of it, and many healthcare providers also have limited knowledge of the disorder. This often forces the caregiver into becoming the “expert” on the disorder when interacting with other family members, friends, and educators. In addition, they become the de facto treatment advocates for their loved ones when attending healthcare appointments and during hospitalizations, in addition to providing care on a daily basis. (We discuss this at greater length in Chapter 3: “Establishing a Relationship with a Mental Healthcare Provider” and Chapter 13: “A Caregiver’s Perspective.”) Caregivers who are not medically trained can feel unprepared to manage complex medical tasks and can feel inadequately supported and unrecognized by members of their loved one’s professional care team.

The caregiver burden can have devastating effects on the caregiver. There is loss of income and decreased socializing as the caregiver reallocates time for caregiving activities, leading to significant changes in lifestyle. Physical effects of caregiver burden include caregiver weight loss, a decrease in self-care and self-health, and sleep deprivation. [8,9,10] Caregiver burden increases the risk of depression and anxiety in caregivers. [11,12] Caregiver burden is also associated with an increased risk of death for caregivers, including a greater risk of death by suicide. [2,13]

While not all caregivers will experience caregiver burden, research has found common risk factors that can increase the likelihood of developing caregiver burden. Demographic risk factors for developing caregiver burden include female gender, low education, low monthly income, and living with the care recipient. [2,14–22] Psychosocial factors for developing caregiver burden include poor psychological health and poorly perceived well-being of the caregiver, caregiver depression and/or anxiety, perceived patient distress, poor coping skills, and increased social isolation and decreased social activity. [2,11,14,17,22–27]

There are also risk factors associated with the context in which caregiving is provided, including the duration of time spent on caregiving, financial distress, and lack of choice in becoming a caregiver. Caregivers on average spend 24 hours a week providing care, with 21% of caregivers spending more than 40 hours a week providing care. [5] As time spent caregiving increases, so does the risk of caregiver burden. [2] Additionally, as many caregivers are uncompensated or under-compensated for their caregiving work, they report spending money out of pocket for caregiving expenses, which can add to financial hardship. [9] Finally, caregivers who feel that they were forced into the role are at higher risk for developing caregiver burden. [28]

**Caregiver Burnout**

At times, caregivers can find themselves overwhelmed by their responsibilities and the additional stressors due to caregiving to the point that they may resent taking care of their loved ones. The caregiver may fantasize about what life would be like without their burden. That in turn can lead to intense feelings of guilt and shame. After all, it isn’t their loved one’s fault that they need so much care and supervision. These emotions are natural and understandable and usually they are transient, triggered by an event or incident outside of the caregiver’s control. When these negative emotions become persistent and interfere with a caregiver’s ability to take care of themselves and/or their loved one, the caregiver is at risk for caregiver burnout.
The term “burnout” can be traced back to an article published in 1974 by psychologist Herbert J. Freudenberger in which he described the emotional and physical exhaustion that he began to experience while engaged in treating his patients. Christina Maslach further outlined three core components of burnout: emotional exhaustion, depersonalization, and reduced personal accomplishments. Emotional exhaustion occurs when an individual feels overextended emotionally or worn out, usually as a result of a build-up of stressors over which the person feels they have no control. The person experiencing emotional exhaustion can feel trapped or stuck in the situation that is causing them to be stressed. Depersonalization is often a response to emotional exhaustion. A person experiencing depersonalization has become emotionally detached from the people around them, including loved ones. The third component, a decline in personal accomplishments, refers to a reduced sense of self-efficacy and a growing sense of inadequacy and inability to cope with the demands being made on the person. In this context, burnout is related to both personal stressors, complex interpersonal relationships, and self-evaluation.

Maslach and colleagues identified six “mismatches” that occur and contribute to burnout: workload, control, reward, community, fairness, and values. A mismatch in any of these areas can lead to burnout. While these principles were initially applied to burnout in the workforce, they also apply to burnout in caregiving.

**Workload.** A workload mismatch occurs when the caregiver believes that it is excessive, and the demands being made on the caregiver lead to exhaustion. However, a caregiver who believes that they lack the skills to be providing the care required by their loved one can also experience a workload mismatch, even if the time needed for the required care is reasonable. For example, a caregiver who is required to clean their loved one’s wound or tracheostomy may feel that they have not received adequate education to complete that task. Ways in which a caregiver can reduce a workload imbalance include asking for help in providing care or delegating other household tasks to other family members. Hiring respite care or household help can also alleviate workload imbalance. If those options are not possible, volunteer help may be available from local charities or religious organizations. It is also important that a caregiver seek out education for any of the tasks they may not feel competent in completing. Ask healthcare providers if they have any educational materials they can share, or if they would demonstrate the techniques they use.

**Control.** A caregiver who feels that they have little control over their resources, such as time or money, can develop a sense of inefficacy. For example, a caregiver who works outside of the home may feel that they are unable to spend adequate time providing care to their loved one, or they may feel guilty over the time they spend away from their loved one. Alternatively, a caregiver who has no other source of income may find little money left over for discretionary spending after paying for bills, healthcare, and groceries, or may need to go without some essentials in order to make ends meet. When it comes to a sense of control, it is important to consider what is within our control and what is not. When we focus on the things we can control, it leads to a sense of self-efficacy. For example, we may not be able to control the amount of time we have to spend on providing care to our loved one, but while caregiving, we can focus on the work we are doing and the positive impact it has on the quality of our loved one’s life.

**Reward.** When it comes to informal caregiving, much of the (sense of) reward is intrinsic. That is, the sense of purpose of providing care to a loved one can be rewarding in itself. However, a caregiver who provides care simply because it is what is expected of them may not feel any great sense of accomplishment from their caregiving. This can be exacerbated if the