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Clinical Nursing Management and Care Considerations in Prader-Willi Syndrome

Horyah Badi Duij Almutiry [(1)] , Salha Shahi Mufdi Alanzi [(2)] , Dalal Khudhayer Assri Alanazi [(2)] , Nawal Saleh Dasan Alharbi [(3)] , Dalal Lafi Almutrafi [(4)] , Saad Abdullah Alwazrah [(5)] , Amjad Saleh Alghamdi [(6)] , Amani Ali Yahya Ghazwni [(7)] , Ebtisam Ibrahim Ali [(8)] , Reem Mohammad Kwes [(9)] , Awd Maud Saad Reshd [(10)] , Alaa Mohammed Almarwani [(11)]

(1) Al Rawdah 1 Health Center – Riyadh,Ministry of Health, Saudi Arabia,
(2) Al Khaleej 2 Health Center,Ministry of Health, Saudi Arabia,
(3) Maternity and Children's Hospital – Hafar Al Batin,Ministry of Health, Saudi Arabia,
(4) King Khalid Hospital – Alkharj,Ministry of Health, Saudi Arabia,
(5) Wadi Dawaser General Hospital – Wadi Dawaser,Ministry of Health, Saudi Arabia,
(6) King Fahd Medical City – Riyadh,Ministry of Health, Saudi Arabia,
(7) Tabuk Health Cluster – King Khalid Hospital,Ministry of Health, Saudi Arabia,
(8) Dar Al-Bayda 1 Health Center – Riyadh,Ministry of Health, Saudi Arabia,
(9) Maternity and Children Hospital – Dammam, Eastern Region,Ministry of Health, Saudi Arabia,
(10) Eradah Mental Health Complex – Hail,Ministry of Health, Saudi Arabia,
(11) Umluj General Hospital, Ministry of Health, Saudi Arabia

Abstract

Background: Prader–Willi syndrome (PWS) is a rare genetic disorder caused by loss of expression of paternally inherited genes on chromosome 15q11.2–q13. It is characterized by multisystem involvement, including neurodevelopmental delay, endocrine dysfunction, hyperphagia, and severe obesity, leading to significant morbidity and mortality.

Aim: This article aims to review the clinical features, pathophysiology, evaluation, and comprehensive nursing management strategies for individuals with Prader–Willi syndrome, with emphasis on multidisciplinary care.

Methods: A narrative review of clinical, genetic, and nursing literature was conducted focusing on etiology, epidemiology, diagnosis, treatment, complications, and patient education related to PWS.

Results: PWS manifests with hypotonia and feeding difficulties in infancy, progressing to hyperphagia, obesity, endocrine abnormalities, behavioral challenges, and cognitive impairment. Accurate diagnosis relies on DNA methylation analysis, while management requires lifelong, coordinated interventions including growth hormone therapy, dietary control, behavioral support, and monitoring of obesity-related complications.

Conclusion: Early diagnosis and sustained multidisciplinary management are critical to improving functional outcomes, reducing complications, and enhancing quality of life in individuals with Prader–Willi syndrome. Nursing professionals play a vital role in education, monitoring, and care coordination.

Key Words: Prader–Willi syndrome; hyperphagia; obesity; genetic disorder; nursing management.

Introduction

Prader-Willi syndrome (PWS) is a rare and multifaceted genetic disorder with significant implications for metabolic, endocrine, and neurological function. It is recognized as the most common syndromic cause of obesity, with its manifestations spanning multiple physiological and behavioral domains. Early life is marked by profound hypotonia, poor muscle tone, and feeding difficulties, often necessitating specialized nutritional support and close monitoring to prevent malnutrition. As children with PWS grow, global developmental delays become evident, affecting motor skills, cognitive function, and speech. By approximately three years of age, hyperphagia emerges, accompanied by excessive weight gain and early-onset obesity, which contribute to long-term metabolic complications. Distinctive physical characteristics include facial dysmorphisms, such as a narrow forehead, almond-shaped eyes, and a thin upper lip, as well as strabismus and musculoskeletal abnormalities, including scoliosis and small hands and feet. These features, alongside behavioral challenges such as temper outbursts, compulsivity, and social difficulties, require comprehensive and ongoing clinical assessment [1]. Hypothalamic dysfunction is central to the pathophysiology of PWS and underlies many of its clinical manifestations. Endocrine abnormalities commonly include growth hormone deficiency, resulting in short stature, reduced lean body mass, and low bone mineral density. Hypogonadism is prevalent, contributing to delayed or incomplete pubertal development and infertility. Other endocrinopathies include hypothyroidism and central adrenal

*Corresponding author e-mail: hbalmutiry@moh.gov.sa (Horyah Badi Duij Almutiry).
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insufficiency, which necessitate careful evaluation and hormone replacement when indicated. The constellation of neurological, metabolic, and endocrine disturbances necessitates a multidisciplinary approach that integrates medical, nutritional, behavioral, and rehabilitative interventions. Genetically, PWS results from the loss of expression of paternally inherited genes on chromosome 15q11.2–q13. This occurs most frequently through paternal deletion, maternal uniparental disomy, or imprinting defects. Recognition of the genetic basis of the syndrome enables confirmatory testing, early diagnosis, and tailored management. In infancy, early identification of hypotonia and feeding challenges permits timely interventions to improve growth and development. In older children, prevention and management of obesity through controlled diet, structured physical activity, and behavioral strategies are critical. Long-term outcomes are enhanced by coordinated care involving endocrinologists, nutritionists, geneticists, psychologists, and nursing professionals, emphasizing the importance of interprofessional collaboration in optimizing health, functional capacity, and quality of life for individuals with PWS [1][2].

Etiology

Prader-Willi syndrome (PWS) arises from the loss of expression of paternally inherited genes located on chromosome 15q11.2–q13. The majority of cases, approximately 70 percent, result from a paternal deletion within this chromosomal region, representing errors in genomic imprinting that disrupt normal gene expression [1][2]. Maternal uniparental disomy accounts for roughly 25 percent of cases, whereby both copies of the relevant chromosomal segment are inherited from the mother, leading to a functional absence of paternally expressed genes. Less commonly, PWS originates from defects in the imprinting center itself, including microdeletions or epimutations, which interfere with the normal regulation of gene activity on chromosome 15 [1][2]. While most instances of PWS are sporadic and arise de novo, familial transmission is occasionally observed. In these cases, a microdeletion within the paternal imprinting center may be inherited from the paternal grandmother, predisposing subsequent generations to the syndrome. Notably, PWS represents the first human disorder definitively linked to genomic imprinting, a phenomenon in which the phenotypic effect of a gene is contingent on its parental origin. This discovery underscored the critical role of epigenetic regulation in human development and disease, highlighting that the expression of specific genes is not solely determined by their sequence but also by the parental source of inheritance [1][2]. Understanding these genetic mechanisms is fundamental to accurate diagnosis, counseling, and targeted management of PWS, as the underlying etiology directly influences clinical presentation, risk assessment, and the potential for recurrence in families with imprinting defects.

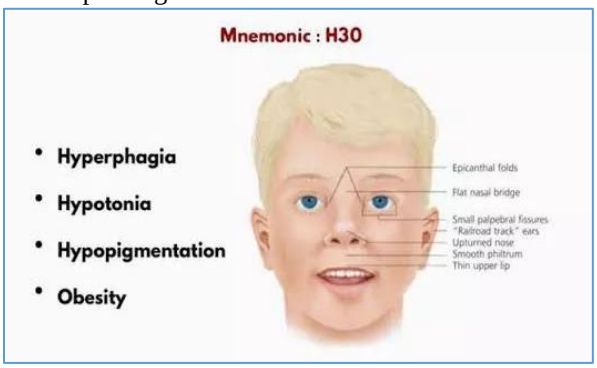

Fig. 1: Parder-Willi Syndrome.

Epidemiology

Prader-Willi syndrome is an uncommon genetic condition with an estimated prevalence ranging from one in 20,000 to one in 30,000 live births [3]. Globally, it is estimated that approximately 400,000 individuals are affected, with roughly 20,000 cases identified in the United States [2]. Despite its relative rarity, PWS is recognized as the most common genetic contributor to severe, life-threatening obesity, reflecting its profound impact on energy balance, satiety regulation, and metabolic control [2]. Epidemiological data indicate that the disorder affects males and females equally, and no significant differences in incidence have been observed across racial or ethnic populations [4]. The early identification of PWS is challenging due to its complex and evolving clinical manifestations. Neonatal hypotonia and feeding difficulties often prompt initial evaluation, but hyperphagia and obesity typically emerge later in childhood, complicating prevalence studies and population-based assessments. Advances in molecular diagnostics, particularly chromosomal microarray analysis and methylation studies, have improved the accuracy of case identification and allowed for more reliable epidemiological estimates. Population-based registries and longitudinal studies have further contributed to understanding the natural history of PWS, the distribution of genetic subtypes, and associated comorbidities. Epidemiological insight is crucial for healthcare planning, resource allocation, and the development of specialized clinical programs, as early intervention and coordinated care can mitigate the long-term consequences of obesity, endocrine dysfunction, and behavioral challenges associated with PWS [2][3][4].

Pathophysiology

The clinical manifestations of Prader-Willi syndrome are fundamentally linked to hypothalamic dysfunction arising from the absence of expression of multiple genes on chromosome 15q11.2–q13. This genetic disruption impairs neuroendocrine regulation, producing hallmark features such as neonatal hypotonia, poor feeding in infancy, hyperphagia, and severe obesity during later childhood and adulthood

[5]. Impairment of hypothalamic neuropeptide signaling compromises satiety mechanisms, reduces energy expenditure, and alters multiple endocrine axes. Consequently, individuals with PWS frequently exhibit growth hormone deficiency, hypogonadism, hypothyroidism, and, in rare instances, central adrenal insufficiency, all of which contribute to abnormal growth, reduced lean body mass, delayed puberty, and increased metabolic risk [5]. Dysregulation of the oxytocin and ghrelin systems plays a significant role in hyperphagia and compulsive food-seeking behavior, further exacerbating obesity and associated metabolic complications. Cognitive and behavioral deficits in PWS have been linked to structural and functional abnormalities in the central nervous system, particularly within the limbic system and frontal cortical pathways. Neuroimaging studies consistently demonstrate reduced volumes of the thalamus, amygdala, and brainstem, correlating with impaired cognitive performance, emotional regulation difficulties, and behavioral dyscontrol [6]. These neuroanatomical changes provide a mechanistic basis for the observed deficits in executive function, social cognition, and adaptive behavior, and they underscore the multifactorial pathophysiology of PWS, which integrates genetic, endocrine, metabolic, and neurological dysfunctions. Understanding these interrelated mechanisms informs the development of targeted therapeutic interventions, including growth hormone therapy, nutritional management, behavioral support, and multidisciplinary care strategies designed to mitigate long-term complications and improve quality of life for affected individuals [5][6].

History and Physical

Prader-Willi syndrome (PWS) presents a broad spectrum of clinical manifestations that evolve across different stages of life, reflecting the progressive nature of the disorder. Symptoms may be observable at birth, though many features become more apparent with age. Hypotonia is nearly universal in affected infants and can be identified prenatally through reduced fetal movements and delayed onset of quickening. At birth, these infants often demonstrate growth parameters, including weight, length, and body mass index, that are 15 to 20 percent lower than those of unaffected siblings, indicating impaired intrauterine growth. This decreased prenatal growth, in conjunction with abnormal fetal positioning, contributes to an elevated rate of assisted deliveries, including cesarean sections. Hypotonia is typically accompanied by poor suckling, feeding difficulties, and diminished muscle mass and strength, leading to early weight gain deficiencies and often necessitating specialized feeding strategies or prolonged enteral nutrition [2][7]. Distinctive facial dysmorphisms are characteristic of PWS and include a narrow frontal diameter, almond-shaped palpebral fissures, strabismus, a slender nasal bridge, thin upper vermillion border, and downturned mouth corners, often with enamel hypoplasia. Additional physical features, such as small hands and feet, may not be evident at birth but typically emerge in early childhood [2][7]. Developmental delays are common, particularly in motor function, with early milestones occurring at approximately half the expected rate. Independent walking is typically achieved around 27 months, significantly later than in unaffected children [7][8]. Language development is often delayed, and cognitive impairments, including learning disabilities, typically become apparent by school age [9]. Behavioral challenges are nearly universal and may include anxiety, obsessive-compulsive tendencies, temper outbursts, and self-injurious behaviors [4].

Endocrine dysfunction is a hallmark of PWS. Growth hormone deficiency is the most frequently observed abnormality, resulting in short stature, growth deceleration, and altered body composition. This deficiency is evident in early childhood and often persists into the second decade, manifesting as truncal obesity and a failure to achieve the normal pubertal growth spurt [1][7][10]. Impairment of the growth hormone-insulin-like growth factor 1 axis has been documented in nearly all patients. The characteristic progression of weight gain and hyperphagia occurs in four distinct phases: infancy marked by hypotonia and poor feeding with slow weight gain (birth to 9–15 months); steady growth with typical weight gain rates (9–24 months); early childhood weight gain without increased appetite (2–4.5 years); development of hyperphagia and heightened interest in food (4.5–8 years); followed by progressive hyperphagia, food-seeking behavior, and impaired satiety through adolescence into adulthood, with eventual resolution of insatiable appetite in later adulthood [2][7][11]. Central obesity is a defining feature, contributing to significant morbidity and mortality. In adults, hyperphagia can precipitate life-threatening events, including choking, and extreme food-seeking behaviors, such as consumption of nonfood items, have been reported. Elevated ghrelin levels may partially explain impaired satiety observed in these patients [13][14]. Hypogonadism is nearly universal. Affected males often present with cryptorchidism requiring orchiopexy, scrotal hypoplasia, and penile underdevelopment, leading to difficulties with standing urination. Testosterone therapy can facilitate early functional milestones such as potty training. Pubertal progression is arrested, with testicular failure and persistent small testicular size observed in adulthood. Females commonly exhibit external genital hypoplasia, delayed breast development, and delayed menarche, often not occurring until the second decade of life. Estrogen and luteinizing hormone levels remain low to normal, whereas follicle-stimulating hormone levels are elevated or normal, suggesting combined central and primary gonadal dysfunction [15][16]. Both sexes are generally infertile, although premature pubarche can occur independently of

obesity and insulin resistance. Approximately 14 to 30 percent of individuals demonstrate advanced bone age [1].

Additional clinical features extend beyond musculoskeletal and endocrine abnormalities. Sleep disturbances, thick viscous saliva, reduced pain perception, decreased vomiting reflex, epilepsy, thermoregulatory instability, scoliosis, kyphosis, and osteoporosis are frequently observed [17]. Hypopigmentation of skin, hair, and iris occurs in approximately 30 to 50 percent of cases relative to the familial phenotype. Obesity-related comorbidities further complicate the clinical course and include cardiovascular disease, metabolic-associated steatotic liver disease, dyslipidemia, diabetes mellitus, sleep apnea, and respiratory compromise. Collectively, these features highlight the multisystemic involvement of PWS and underscore the necessity for comprehensive, multidisciplinary assessment and ongoing monitoring to manage its complex medical, developmental, and behavioral sequelae.

Evaluation

The evaluation of suspected Prader-Willi syndrome (PWS) relies primarily on molecular genetic testing, as clinical features alone may be insufficient for definitive diagnosis, particularly in infancy. DNA methylation analysis is considered the gold standard, capable of detecting more than 99 percent of PWS cases [18]. A positive methylation result confirms the diagnosis of PWS but does not specify the underlying genetic mechanism. Therefore, subsequent testing is required to differentiate between paternal deletion, maternal uniparental disomy, or imprinting defects. Fluorescence in situ hybridization (FISH) is commonly employed to detect deletions within the 15q11–q13 region, while chromosomal microarray analysis allows identification of maternal disomy and other subtle chromosomal abnormalities [2][19]. In settings with advanced diagnostic resources, genetic confirmation is often achieved within the first few months of life, sometimes as early as two months. However, in many cases, the definitive diagnosis is delayed until approximately four years of age, when more distinctive clinical features, including hyperphagia, behavioral abnormalities, and characteristic facial dysmorphisms, become apparent [9]. Beyond molecular confirmation, evaluation should include assessments tailored to the multisystem involvement of PWS. Endocrine evaluation is critical, as hypothyroidism and growth hormone deficiency are common. Thyroid function tests are recommended prior to initiating growth hormone therapy to ensure optimal treatment safety and efficacy [2]. Serum levels of insulin-like growth factor 1 and its binding protein, IGFBP-3, provide additional information on growth hormone status and help guide supplementation strategies [2]. Metabolic screening is indicated for obesity-related complications. Fasting glucose, hemoglobin A1c, and oral glucose tolerance testing are advised when there is suspicion of insulin resistance or diabetes mellitus, conditions frequently observed in this population. Assessment of sleep quality is an important component of PWS evaluation, as sleep-disordered breathing, including obstructive sleep apnea, is highly prevalent. Polysomnography provides objective measurement of sleep patterns, oxygen desaturation, and apnea events. Skeletal assessment should also be incorporated, given the risk of reduced bone mineral density due to growth hormone deficiency, hypogonadism, and decreased physical activity. Dual-energy x-ray absorptiometry (DEXA) is recommended to evaluate bone mineral content and composition, enabling early intervention to prevent osteoporosis and fractures [2]. Collectively, these evaluations provide a comprehensive framework for diagnosis, risk stratification, and individualized management planning, supporting improved long-term outcomes in individuals with PWS.

Treatment / Management

Management of Prader-Willi syndrome (PWS) requires a comprehensive, age-specific, and multidisciplinary approach that addresses the syndrome's multisystemic manifestations, including metabolic, endocrine, musculoskeletal, cognitive, and behavioral disturbances. Care strategies vary depending on the developmental stage, starting from infancy and continuing into adulthood, with interventions tailored to the evolving clinical features. Early recognition of hypotonia, feeding difficulties, and delayed growth in infants is critical for initiating supportive care and preventing complications associated with undernutrition and developmental delays. Feeding teams often assess infants to implement specialized techniques that compensate for poor sucking reflexes and low muscle tone. High-calorie formulas or supplements may be necessary to achieve adequate weight gain, particularly in cases of severe hypotonia and failure to thrive [9][20]. As children age and hyperphagia emerges, dietary management becomes central to preventing severe obesity, a hallmark of PWS that significantly increases morbidity and mortality. Most children with PWS require approximately 70 percent fewer calories than age-matched peers due to decreased basal energy expenditure and impaired satiety mechanisms. Strict caloric restriction must be carefully balanced to ensure adequate intake of essential vitamins and minerals. Nutritional supplementation is often necessary, but products such as gummy vitamins must be used with caution because of their caloric content and potential for accidental overconsumption, especially given the disorder's characteristic compulsive eating behaviors. Caregivers are encouraged to implement physical food security measures, including locking refrigerators and cupboards, to reduce the risk of uncontrolled access. Training in emergency procedures, including the Heimlich maneuver, is essential, as individuals with PWS often do not chew thoroughly and are at heightened risk of choking. For older children and adults with severe obesity, pharmacologic

interventions and, in select cases, bariatric surgery may provide additional tools for weight management [9].

Physical and occupational therapy play a crucial role in addressing hypotonia and motor delays, enhancing strength, coordination, and overall functional independence. Growth hormone therapy with recombinant human growth hormone (rhGH) is a cornerstone of treatment and is recommended as early as possible, ideally before the first birthday. Early initiation of rhGH therapy improves muscle tone, strength, and physical function, in addition to promoting linear growth. While formal growth hormone stimulation testing is not mandatory for treatment initiation, ongoing evaluation of growth parameters and endocrine status is necessary. Continuation of rhGH therapy into adolescence maximizes final height outcomes and contributes to increased lean body mass, reduced fat mass, improved bone mineral density, and enhanced motor function. Although daily or weekly injections are required, long-term benefits, including improved stature and metabolic profile, justify routine administration [10]. Adults with persistent growth hormone deficiency may also derive metabolic and functional benefits from continued rhGH therapy. Endocrine management extends beyond growth hormone replacement. Cryptorchidism is observed in approximately 50 percent of male infants with PWS, frequently requiring orchiopexy, although human chorionic gonadotropin therapy may be beneficial in select cases. At puberty, hormonal replacement is often indicated due to hypogonadism. Boys generally receive testosterone therapy to promote secondary sexual characteristics, support pubertal development, and facilitate functions such as potty training by increasing penile size. Girls may require estrogen supplementation via transdermal patches to initiate and maintain secondary sexual characteristics and menstrual function. Pubertal progression is commonly delayed, and spontaneous fertility is rare in both sexes. Monitoring of gonadal function, puberty, and bone health is essential, with endocrinology consultation recommended for individualized management [1][7].

Behavioral and psychiatric manifestations are prominent throughout childhood and adulthood. Temper outbursts, obsessive–compulsive behaviors, rigidity, skin picking, and mood or psychotic disorders are frequently reported. Approximately 25 percent of children with PWS meet criteria for autism spectrum disorder. Behavioral interventions are critical to minimize maladaptive behaviors, help patients understand structured routines, and support compliance with dietary restrictions. Pharmacologic management may be warranted for severe mood or psychotic symptoms. Skin picking, a common compulsive behavior, has demonstrated responsiveness to oral guanfacine in some studies, although behavioral modification remains the first-line approach [22]. Cognitive impairments affect academic achievement, vocational independence, and social integration, requiring individualized educational plans during childhood and structured vocational or supervised settings for adults. Accelerated cognitive decline in older adults highlights the need for ongoing neuropsychological monitoring [21]. Skeletal and musculoskeletal health requires periodic evaluation. Reduced bone mineral density is common due to hypogonadism, growth hormone deficiency, and decreased physical activity. Dual-energy x-ray absorptiometry (DEXA) scans are recommended every 2 to 3 years starting at approximately five years of age. Scoliosis and kyphosis are frequent complications, necessitating routine orthopedic assessment and timely interventions when indicated. Routine thyroid function testing should commence during the first year of life to detect hypothyroidism, which may exacerbate growth and metabolic abnormalities [23]. Obesity-related complications, including type 2 diabetes, dyslipidemia, metabolic-associated steatotic liver disease, cardiovascular disease, and respiratory dysfunction, require ongoing laboratory and clinical monitoring. Sleep-disordered breathing, including obstructive sleep apnea, affects most pediatric and young adult patients with PWS. Clinical suspicion should be raised in individuals exhibiting daytime sleepiness, behavioral changes, or poor concentration, with polysomnography recommended for diagnosis. Surgical interventions, including tonsillectomy or adenotonsillectomy, may be necessary in select cases to alleviate upper airway obstruction.

Until recently, treatment options for PWS were supportive rather than disease-specific. In March 2025, the Food and Drug Administration (FDA) approved VYKAT XR (diazoxide choline) extended-release tablets for hyperphagia management in patients aged four years and older. Clinical trials conducted prior to approval demonstrated the medication’s efficacy in reducing food-seeking behaviors and improving satiety, providing a pharmacologic option to complement behavioral and dietary interventions [24][25]. Overall, management of PWS requires coordinated care from an interprofessional team that includes pediatricians, endocrinologists, nutritionists, occupational and physical therapists, psychologists, behavioral therapists, and genetic counselors. This comprehensive approach addresses the multisystemic nature of the syndrome, mitigates complications, improves functional outcomes, and enhances quality of life across the lifespan. Early identification, timely interventions, and consistent monitoring are essential to optimize long-term prognosis and prevent the morbidity and mortality associated with obesity, endocrine dysfunction, and neurobehavioral complications in individuals with PWS.

Differential Diagnosis

The clinical features of Prader-Willi syndrome (PWS) can overlap with those of several other disorders, necessitating careful differential diagnosis to ensure accurate identification and timely management. Neonatal hypotonia, a hallmark of PWS, is also observed in various neuromuscular and metabolic disorders. For example, spinal muscular atrophy and congenital myopathies such as myotonic dystrophy may present with poor respiratory effort, generalized hypotonia, and feeding difficulties during early life [7]. These conditions, while sharing certain phenotypic traits with PWS, differ in genetic etiology, progression, and associated comorbidities. A group of disorders termed Prader-Willi-like syndromes may also mimic PWS, presenting with hypotonia, short stature, obesity, and developmental delays. However, these syndromes involve distinct chromosomal abnormalities or gene mutations that do not include the paternal 15q11–q13 deletion or imprinting defects characteristic of PWS [26]. CNS tumors affecting hypothalamic function, particularly craniopharyngioma, may induce features similar to PWS, including impaired growth, obesity, and endocrine dysfunction. The overlap in clinical presentation arises primarily from the impact of hypothalamic injury or tumor-related therapies on appetite regulation, growth hormone secretion, and pubertal development [7]. Despite these similarities, the progressive, age-dependent pattern of PWS—beginning with hypotonia and feeding difficulties in infancy, followed by the emergence of hyperphagia and subsequent severe obesity—is unique. This temporal evolution of symptoms provides a distinguishing feature that facilitates differentiation from other genetic or acquired disorders. Accurate genetic testing remains the cornerstone of diagnosis, confirming the presence of the specific paternal deletion, maternal uniparental disomy, or imprinting defect that defines PWS. Recognizing the subtle distinctions between PWS and other hypotonic or obesity-related syndromes is essential for targeted management, early intervention, and prevention of long-term complications associated with the disorder.

Prognosis

The prognosis for individuals with Prader-Willi syndrome is variable and closely linked to the timing of diagnosis, implementation of early interventions, and the management of obesity and associated comorbidities. Early recognition and treatment significantly improve outcomes, particularly by mitigating the progression of hyperphagia and preventing severe obesity, which is a major contributor to morbidity and mortality [2][27]. With appropriate interventions—including dietary control, growth hormone therapy, endocrine management, and behavioral support—affected individuals can achieve improved physical growth, metabolic regulation, and cognitive development. However, intellectual disability and behavioral challenges typically persist, limiting the potential for full independence in adulthood. Consequently, most individuals with PWS require lifelong supportive services, including assisted living arrangements or supervised vocational programs, to maintain safety, health, and quality of life. Mortality in PWS is frequently associated with obesity-related complications. Cardiovascular disease, type 2 diabetes mellitus, respiratory insufficiency, and gastrointestinal emergencies are common causes of death, often occurring in the fourth decade of life when obesity and metabolic dysfunction are poorly managed. In contrast, individuals who maintain a controlled weight and adhere to structured dietary, endocrine, and physical activity interventions have demonstrated the capacity to achieve extended life expectancy, reaching into the seventh decade or beyond. Lifelong monitoring, family education, and adherence to multidisciplinary care protocols are essential to optimize longevity and reduce preventable complications. Despite these advances, the prognosis remains guarded for those with delayed diagnosis, inadequate nutritional control, or unmanaged endocrine and behavioral disturbances. The integration of early intervention programs, endocrine therapy, and comprehensive medical and psychosocial support is critical in shaping a more favorable long-term outcome for patients with PWS.

Complications

Complications of Prader-Willi syndrome are predominantly linked to obesity and its systemic consequences, although additional issues reflect musculoskeletal, neurologic, endocrine, and behavioral dysfunction. Obesity is central to increased morbidity, precipitating metabolic syndrome, type 2 diabetes, dyslipidemia, and cardiovascular disease. Respiratory complications, including obstructive sleep apnea, hypoventilation, obesity-related respiratory failure, and recurrent aspiration pneumonia, are common, particularly in individuals with severe obesity [28]. Cardiovascular sequelae include hypertension and right-sided heart failure, often secondary to chronic hypoxia and pulmonary complications. Gastrointestinal issues arise from delayed gastric emptying, gastroparesis, metabolic dysfunction-associated steatotic liver disease, and the risk of gastric perforation or obstruction due to compulsive binge eating behaviors [29]. Musculoskeletal complications are also significant, with osteoporosis, scoliosis, kyphosis, and hip dysplasia frequently observed. The combination of hypotonia, reduced physical activity, and hormonal deficiencies contributes to decreased bone mineral density, increasing the risk of fractures and deformities. Dermatologic issues, including stasis ulcers and recurrent cellulitis, often result from poor circulation, immobility, and obesity-related pressure changes. Behavioral and cognitive manifestations further compound complications. Self-injurious behaviors, such as skin picking, pose a risk for infection and physical injury while also increasing caregiver burden and decreasing overall quality of life.

Seizures, temperature dysregulation, and persistent hypotonia reflect the neurologic involvement inherent to PWS [2]. Endocrine complications, such as type 2 diabetes mellitus and adrenal insufficiency, are prevalent, particularly in the context of obesity, growth hormone deficiency, and hypogonadism [30]. Early recognition, vigilant monitoring, and proactive management of these complications are critical to reducing morbidity, preventing irreversible damage, and improving both functional outcomes and life expectancy.

Patient Education

Preventive strategies and patient education form an integral part of PWS management. Clinicians should maintain heightened awareness for early signs in infants, including hypotonia, poor sucking reflex, slow weight gain, and cryptorchidism, to facilitate timely diagnosis. Early identification allows the initiation of interventions that mitigate complications and support growth and development. Family caregivers should receive education about the natural progression of PWS, including the predictable transition from early hypotonia and feeding difficulties to hyperphagia, behavioral challenges, and the onset of obesity. Training caregivers in dietary management, behavioral reinforcement strategies, and safe feeding techniques is crucial to prevent serious sequelae associated with uncontrolled appetite and obesity. Lifelong adherence to structured nutritional plans, behavioral interventions, and comprehensive medical and educational support is necessary to minimize morbidity, promote functional independence, and enhance quality of life [30].

Other Issues

Prader-Willi syndrome is caused by the absence of expression of paternally inherited genes on chromosome 15q11–q13, typically due to deletion, maternal uniparental disomy, or imprinting defects. Clinically, it is characterized by neonatal hypotonia, poor feeding, developmental delays, hyperphagia, and the subsequent development of central obesity. Distinctive facial features, such as almond-shaped eyes and a thin upper lip, are often present. DNA methylation analysis provides a highly sensitive diagnostic tool, detecting abnormalities in over 99 percent of cases. Hypotonia and feeding difficulties dominate infancy, while hyperphagia emerges around age three, often accompanied by behavioral challenges including food-seeking behaviors. Fertility is generally absent, and central obesity contributes significantly to morbidity and mortality, emphasizing the importance of early intervention and structured care [2][7].

Enhancing Healthcare Team Outcomes

Optimal care for individuals with PWS requires a coordinated interprofessional approach. Healthcare providers should maintain vigilance for hypotonia, feeding difficulties, slow growth, and emerging hyperphagia to ensure timely diagnosis and intervention. Upon diagnosis, collaboration among genetics specialists, endocrinologists, and developmental medicine professionals is critical for evaluating and managing multisystem complications. Genetic counseling for parents allows assessment of recurrence risk in future pregnancies. Nutritionists and dietitians play a central role in guiding caloric intake, ensuring growth, and mitigating obesity-related complications, while feeding teams support weight gain and safe feeding practices in infancy. Early intervention programs address developmental delays, including physical, occupational, and speech therapy. School-aged children benefit from individualized educational programs that incorporate therapies tailored to cognitive, behavioral, and motor deficits. Clinical pharmacists contribute by optimizing dosing of hormonal therapies and other medications. Nursing staff monitor treatment responses, growth, and laboratory parameters, coordinating care and providing ongoing education to families regarding dietary, behavioral, and therapeutic needs. Mental health nurses, psychiatrists, and psychiatric nurse practitioners manage mood disorders, obsessive-compulsive behaviors, and psychosis. Home health nurses assist families with daily strategies to manage food-seeking behaviors safely. Orthopedic and rehabilitation specialists support functional mobility, while interprofessional coordination ensures comprehensive monitoring and early intervention for complications. This team-based approach optimizes clinical outcomes, reduces morbidity, and improves quality of life and longevity for individuals with PWS [29][30].

Conclusion:

Prader–Willi syndrome is a complex, lifelong genetic disorder that significantly affects physical, metabolic, cognitive, and behavioral health. Its multisystem nature demands early recognition and sustained, comprehensive care to reduce morbidity and improve long-term outcomes. Advances in molecular diagnostics have enabled earlier and more accurate identification, allowing timely initiation of targeted interventions during infancy and childhood. Effective management focuses on strict nutritional control, growth hormone replacement, endocrine monitoring, behavioral support, and prevention of obesity-related complications. As hyperphagia and behavioral challenges progress with age, structured environments, caregiver education, and close supervision of food access become essential for patient safety and quality of life. Nursing professionals play a central role throughout the lifespan of individuals with Prader–Willi syndrome. Their responsibilities include ongoing clinical monitoring, coordination of multidisciplinary care, patient and family education, and early identification of complications such as metabolic disorders, sleep apnea, and musculoskeletal abnormalities. Evidence indicates that individuals who receive early intervention, consistent nutritional

management, and long-term hormonal and behavioral support can achieve improved functional status and extended life expectancy. Ultimately, a coordinated interprofessional approach, with nursing care at its core, is fundamental to enhancing health outcomes, minimizing preventable complications, and supporting individuals with Prader–Willi syndrome and their families across all stages of life.

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