Case Report / *Olgu Sunumu*

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Prader–Willi Syndrome and Obstructive Sleep Apnea Syndrome: A Case Series from Türkiye

Prader–Willi Sendromu ve Obstrüktif Uyku Apne Sendromu: Türkiye'den Bir Olgu Serisi

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Abstract

Prader–Willi syndrome (PWS) is a rare genetic disorder that is frequently associated with sleep-disordered breathing, particularly obstructive sleep apnea syndrome (OSAS). We present a case series of five Turkish children with PWS who underwent overnight polysomnography. OSAS was identified in three patients (60%), one of whom had severe disease. Ear, nose, and throat evaluations revealed adenoid hypertrophy in two patients and tonsillar hypertrophy in one child. These findings suggest that OSAS is clinically relevant in PWS, and underscore the importance of routine polysomnographic assessment before and during growth hormone therapy.

Keywords: Prader-Willi syndrome, obstructive sleep apnea syndrome, polysomnography, growth hormone

Öz

Prader–Willi sendromu (PWS), özellikle obstrüktif uyku apne sendromu (OSAS) ile ilişkili olan nadir bir genetik bozukluktur. Bu çalışmada, gece boyunca polisomnografi uygulanan beş Türk PWS'li çocuğa ait bir olgu serisini sunuyoruz. Beş hastanın üçünde (%60) OSAS saptandı; bunlardan birinde hastalık ağır düzeydeydi. Kulak, burun ve boğaz değerlendirmelerinde iki çocukta adenoid hipertrofisi, bir çocukta ise tonsil hipertrofisi tespit edildi. Bu bulgular, OSAS'ın PWS'de klinik açıdan önemli olduğunu göstermekte ve büyüme hormonu tedavisinden önce ve tedavi sırasında rutin polisomnografik değerlendirmenin önemini vurgulamaktadır.

Anahtar Kelimeler: Prader–Willi sendromu, tıkayıcı uyku apne sendromu, polisomnografi, büyüme hormonu

Introduction

Prader–Willi syndrome (PWS) is a rare neurodevelopmental disorder that occurs due to the loss of paternally inherited genes on chromosome 15q11–q13. Its core features are hypotonia, hyperphagia, short stature and cognitive impairment (1), along with excessive daytime sleepiness (EDS) that reflects frequent sleep–disordered breathing (SDB) (2). Obstructive sleep apnea syndrome (OSAS), a form of SDB, is associated with significant morbidity and worsens the quality of life (3). OSAS is recurrent upper airway obstruction that occurs during sleep, and causes intermittent hypoxemia, hypercapnia, and sleep fragmentation (4). The prevalence of OSAS in children is 1–4% and linked to cardiovascular, metabolic, and neurocognitive consequences (5). It is also highly prevalent in PWS patients due to obesity, craniofacial anomalies, hypotonia, and adenotonsillar hypertrophy (3,6). While there is substantial data on PWS from international studies, Turkish reports are scarce. Herein, we present the cases of five children with PWS undergoing polysomnography (PSG), with focus on the prevalence, ear, nose and throat (ENT) findings, and clinical implications.

Case Report

Children with genetically or clinically confirmed PWS who underwent overnight PSG in our sleep laboratory between 2018 and 2025 were included in the study. The exclusion criteria were

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the lack of confirmed PWS diagnosis, incomplete PSG data, or missing ENT evaluation. The indications were snoring, EDS, or pre–growth hormone (GH) assessment. PSGs were scored using the Embla system per American Academy of Sleep Medicine pediatric criteria (4). The apnea–hypopnea index (AHI), oxygen saturation, and arousal index were evaluated. The severity of OSAS was categorized as normal (AHI <1), mild (1-5), moderate (5-10), and severe (>10). Indirect ENT examinations were performed by a pediatric otolaryngologist, and the adenoid and tonsil size were recorded. Written informed consent was obtained from the parents or legal guardians of all children, and the study was conducted in accordance with the principles of the Declaration of Helsinki (2013 revision).

Five children, including three males and two females (mean age 6.8 ± 5 years), were enrolled in the study. The mean body mass index was 19.6 ± 9.2 kg/m^2. Neonatal hypotonia was common, and craniofacial anomalies and obesity were also noted. ENT evaluation revealed adenoid hypertrophy in two children and tonsillar hypertrophy in one (Table 1). The mean AHI was 10.5 ± 17.2 events/h, and three patients (60%) had OSAS. Minimum oxygen saturation ranged from 72–84% (Table 2).

Discussion

SBD is a major cause of morbidity in patients with PWS, and is driven by hypotonia, craniofacial anomalies, and obesity (3,6). Disrupted sleep worsens neurocognitive, behavioral, and cardiometabolic outcomes (2,5,7). Over half of children with PWS have OSAS, often moderate to severe, leading to EDS, cardiovascular risk, and reduced quality of life (5-7). Other sleep disorders, such as central apnea, narcolepsy–like sleepiness, insomnia, and periodic limb movements, are related to hypothalamic dysfunction (8,9). These mechanisms may explain persistent sleepiness despite OSAS treatment, thereby underscoring the need for comprehensive sleep evaluation.

Three of the five patients (60%) in this study had OSAS, including one severe case. This proportion falls within international ranges, although severe cases may reflect small sample size, referral bias, or variability in obesity, hypotonia, or craniofacial features. ENT evaluation identified upper–airway hypertrophy in three patients, which is consistent with the involvement of adenotonsillar obstruction but also indicates that anatomical factors alone do not explain OSAS severity in PWS patients (6). The patient with severe OSAS was obese, which supports obesity as an additional risk factor. Moreover, age–related differences may also contribute, as younger children tend to present with adenotonsillar obstruction, whereas obesity and hypotonia become more prominent determinants with increasing age.

PSG showed reduced rapid–eye movement (REM) sleep and frequent arousals. Likewise, previous studies have reported

Table 1. Clinical, genetic, AHI, and ENT findings in children with PWS.

Case	Age (years)	Sex	BMI (kg/m^2)	Symptoms	Genetics	AHI (events/h)	ENT findings	Treatment
1	5	F	13.8	Snoring, EDS, apnea	Negative	0.9	Tonsillar hypertrophy, Grade 3	Follow-up
2	5	F	13.9	Snoring, EDS, apnea, neck flexure (ESAP)	Chromosome 15q11–13 deletion	0.2	Adenoid hypertrophy, Grade 3	Follow-up
3	8	M	32.9	Hypotonic birth, snoring	Chromosome 15: 11–13q deletion	41.7	Micrognathia, Mallampati Class IV	CPAP 7 mbar
4	1	M	16.4	EDS, apnea, pre-GH control	Negative	8.7	Adenoid vegetation, adenoid hypertrophy, Grade 3	CPAP 6 mbar
5	15	M	31.1	Pre-GH control	Chromosome 15: 11–13q deletion	2.9	Normal	CPAP 5 mbar

AHI: Apnea–hypopnea index, BMI: Body mass index, CPAP: Continuous positive airway pressure, EDS: Excessive daytime sleepiness, ENT: Ear–nose–throat, ESAP: Excessive short/anteriorly positioned neck (short and thick neck morphology predisposing to obstructive sleep apnea syndrome), GH: Growth hormone, F: Female, M: Male, Mallampati classification: Method for airway assessment, PWS: Prader–Willi syndrome.

Table 2. Polysomnographic findings of children with PWS.

Case	TST (min)	Sleep efficiency (%)	REM (%)	NREM (%)	Mean SpO_2 (%)	Min SpO_2 (%)	AHI (events/h)	Arousal index (events/h)
1	462	86.6	13.2	86.8	92	84	0.9	5.8
2	470	97.0	16.7	83.3	96	72	0.1	11.6
3	412	94.6	20.1	79.9	95	72	48.1	66.5
4	337	79.3	33.5	66.5	96	82	8.7	15.0
5	410	77.4	17.0	83.0	95	80	2.9	11.2

TST: Total sleep time, REM: Rapid eye movement, NREM: Non-rapid eye movement, SpO_2: Peripheral oxygen saturation, AHI: Apnea–hypopnea index, PWS: Prader–Willi syndrome.

altered sleep architecture in PWS patients, such as reduced REM and fragmented sleep (2,3). O'Donoghue et al. (6) linked these changes to neurobehavioral problems, and a review confirmed disrupted continuity and reduced REM as a characteristic of PWS (8). Such abnormalities may contribute to persistent EDS and cognitive impairment beyond OSAS.

The relevance of OSAS screening is heightened by GH therapy. Although GH improves stature, body composition, and quality of life, it has been linked to worsening OSAS in children with obesity or adenotonsillar hypertrophy (2,3). Accordingly, PSG prior to and during GH therapy is recommended (7). Our cohort supports this practice, as most patients underwent PSG before GH initiation.

Conclusion

Recently, a study conducted in Türkiye reported PSG–based findings in PWS (10). Our case series contributes to the current evidence, but is limited by sample size and follow–up. Nevertheless, our findings underscore the need for routine PSG and multidisciplinary care for PWS patients, especially with GH therapy.

Ethics

Informed Consent: The written informed consent was obtained from all patients included in the study.

Footnotes

Authorship Contributions

Data Collection or Processing: G.A., Analysis or Interpretation: G.A., T.Y., U.O.A., Literature Search: G.A., T.Y., Writing: G.A., T.Y., U.O.A.

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