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ORIGINAL ARTICLE

Sleep phenotype in children with genetically confirmed achondroplasia: Polysomnographic and neuroradiological findings

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ABSTRACT

Objectives: This study aimed to evaluate polysomnographic (PSG), craniocervical magnetic resonance imaging (MRI), and treatment findings in children with genetically confirmed achondroplasia.

Patients and methods: This retrospective single-center study included 14 children with achondroplasia who underwent overnight PSG between January 2022 and December 2025. Demographic characteristics and PSG and MRI findings were reviewed. Obstructive sleep apnea (OSA) severity was classified using the obstructive apnea-hypopnea index (OAHI), while the total apnea-hypopnea index (AHI) and central apnea index (CAI) were assessed separately.

Results: Based on OAHI classification, five patients (35.7%) had OSA, including three with mild and two with severe disease. Central respiratory events were observed in nine patients (64.3%); however, none fulfilled the diagnostic criteria for central sleep apnea syndrome (CAI ≥ 5 events/h). Median total AHI, REM-AHI, and CAI were 1.06 (0.57-2.93), 2.24 (0.00-7.97), and 0.32 (0.00-0.94) events/h, respectively. Mean sleep efficiency was 90.1%. Magnetic resonance imaging showed foramen magnum (FM) stenosis in seven of 13 patients, five of whom had undergone decompression. Five patients with FM stenosis had central respiratory events, and four had OSA, including two with severe disease. On postoperative PSG, four of five children had total AHI ≥ 1 event/h, while three had OSA (two mild and one severe). None of the six children without FM stenosis had OSA; the remaining child with mild OSA had no available MRI.

Conclusion: Sleep-disordered breathing is common in children with achondroplasia. Central respiratory events and OSA were numerically more frequent in children with FM stenosis; these observations require cautious interpretation in this small cohort. Polysomnography, neuroimaging, and clinical findings should be evaluated together in patients with achondroplasia.

Keywords: Achondroplasia, central respiratory events, foramen magnum stenosis, obstructive sleep apnea, polysomnography.

Achondroplasia is the most common form of skeletal dysplasia and results from autosomal dominant mutations in the FGFR3 (fibroblast growth factor receptor-3) gene. Beyond disproportionate short stature, affected children exhibit characteristic craniofacial and upper airway abnormalities that predispose them to sleep-related breathing disorders (SRBDs), one of the major causes of morbidity in childhood.[1]

Obstructive sleep apnea (OSA) is substantially more common in children with achondroplasia than in the general pediatric population, with reported prevalence rates exceeding 50%.[2] This increased susceptibility is mainly attributed to midface hypoplasia, relative macroglossia, upper airway narrowing, and adenotonsillar hypertrophy.[2] In addition to OSA, children with achondroplasia may develop central sleep apnea

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syndrome (CSAS), most commonly due to foramen magnum (FM) stenosis causing compression of the cervicomedullary junction and brainstem respiratory centers.[2,3] Distinguishing obstructive from central respiratory events is clinically important because their pathophysiology, management, and prognosis differ substantially. Untreated SRBDs have been associated with intermittent hypoxemia, sleep fragmentation, neurocognitive impairment, behavioral problems, learning difficulties, and reduced quality of life.[4,5]

International consensus recommendations advocate routine polysomnography (PSG) surveillance as part of the multidisciplinary management of children with achondroplasia.[6] Polysomnography provides objective characterization of sleep-disordered breathing and allows assessment of respiratory events and associated sleep parameters.[7] Although previous studies have described SRBDs in children with achondroplasia, important aspects such as sleep architecture, central respiratory events, nocturnal oxygenation, and associated neuroradiological findings have not always been evaluated together.[2,8,9] In addition, differences in study design, patient characteristics, and PSG scoring methods have contributed to variability across published findings. Further well-characterized pediatric cohorts may help refine the current understanding of the sleep phenotype of achondroplasia and inform clinical management.

This study aimed to evaluate overnight PSG findings in children with genetically confirmed achondroplasia assessed within a follow-up program and examine sleep architecture, SRBDs, nocturnal oxygenation, and accompanying sleep disorders. This cohort sought to add clinical data on central respiratory events and OSA by presenting each child's PSG findings alongside MRI findings graded using the Achondroplasia Foramen Magnum Score (AFMS) and treatment history.

PATIENTS AND METHODS

This single-center, retrospective observational study included 14 children with achondroplasia who underwent overnight PSG at the Sleep Disorders Center of Çukurova University Faculty of Medicine between January 2022 and December 2025. Subsequent treatment and available follow-up PSG data were updated through September 2026. Patients were referred by the Pediatric Endocrinology Department for systematic evaluation within a multidisciplinary follow-up program regardless of disease severity. Fourteen children with genetically confirmed achondroplasia who completed overnight PSG were included in the study. Demographic characteristics, anthropometric measurements, clinical data, PSG findings, MRI findings, and treatment information, including FM decompression before the first PSG, adenoidectomy before or after the first PSG, and post-PSG positive airway pressure (PAP) therapy, were retrospectively obtained from electronic medical records and the sleep laboratory database. Written informed consent was obtained from all participants. The study protocol was approved by the Research Ethics Committee of the Faculty of Medicine, Çukurova University (Data: March 13, 2026, Approval No: 164). The study was conducted in accordance with the principles of the Declaration of Helsinki.

The following demographic and clinical variables were recorded for each patient: age, sex, height, weight, body mass index (BMI), head circumference, history of surgical interventions, and subsequent PAP therapy.

Total sleep time, sleep efficiency, sleep latency, REM latency, percentages of N1, N2, N3, and REM sleep, total apnea-hypopnea index (AHI), obstructive apnea-hypopnea index (OAHI), REM-AHI, central apnea index (CAI), mean, minimum, and maximum heart rate, mean and minimum oxygen saturation, time spent with peripheral oxygen saturation below 88%, periodic limb movements, and the number of central apneas were evaluated by PSG. The CAI was calculated by dividing the number of central apneas by total sleep time (hours). The presence of central respiratory events was recorded separately from the diagnosis of CSAS.

Available cranial and cervical MRI examinations were reviewed for the presence of FM stenosis, cervicomedullary junction abnormalities, cerebellar tonsillar ectopia, ventricular enlargement, and other associated craniovertebral abnormalities. The severity of FM stenosis was graded using the AFMS, ranging from 0 (no stenosis) to 4 (myelopathic T2 signal changes).[10] Preoperative MRI scans were used for children who had undergone FM decompression.

All participants underwent attended overnight PSG using a Natus system (Natus Medical Inc., Pleasanton, CA, USA). Standard

recordings included electroencephalography, electrooculography, submental and tibial electromyography, electrocardiography, nasal airflow, thoracoabdominal respiratory effort, pulse oximetry, and body position monitoring. Sleep stages and respiratory events were scored according to the pediatric criteria of the American Academy of Sleep Medicine Manual Version 3.0 by an experienced sleep physician.[11] Obstructive sleep apnea severity was classified according to the OAHI using pediatric criteria: OAHI < 1 event/h, normal; 1 ≤ OAHI < 5 events/h, mild OSA; 5 ≤ OAHI < 10 events/h, moderate OSA; and OAHI ≥ 10 events/h, severe OSA.[12] Central respiratory events were assessed using CAI. A CAI < 5 events/h was considered within the normal pediatric range, whereas CAI ≥ 5 events/h was considered diagnostic of CSAS.[13] Total AHI, OAHI, REM-AHI, and CAI were analyzed separately.

Statistical analyses

Statistical analyses were performed using IBM SPSS version 26.0 software (IBM Corp., Armonk, NY, USA). Continuous variables were presented as mean ± standard deviation (SD) or median (interquartile range, IQR), according to data distribution, while categorical variables were expressed as frequencies and percentages. The frequency of central respiratory events in children with and without documented FM stenosis was compared using the two-sided Fisher's exact test due to the small expected cell counts. A *p*-value < 0.05 was considered statistically significant.

RESULTS

The study included 14 children (mean age: 5.5 ± 4.3 years; range, 1 to 14 years) with genetically confirmed achondroplasia, of whom 10 (71.4%) were male. Baseline demographic, anthropometric, and clinical characteristics are presented in Table 1. Three children (21.4%) underwent adenoidectomy during the available observation period: two before the first PSG and one after the first PSG.

Polysomnography findings are presented in Table 2. The mean total sleep time was 393.3 ± 59.6 min, sleep efficiency was 90.1 ± 8.6%, sleep latency was 9.3 ± 7.3 min, and REM latency was 212.0 ± 113.8 min. The mean wake time was 43.7 ± 41.5 min, while mean sleep maintenance was 92.4 ± 8.4%. Two children (14.3%) had BMI values above the 95th percentile on age- and sex-specific BMI charts for children with achondroplasia.

The median total AHI was 1.06 (IQR, 0.57-2.93) events/h, median OAHI was 0.64 (IQR, 0.24-1.84) events/h, median REM-AHI was 2.24 (IQR, 0.00-7.97) events/h, and median CAI was 0.32 (IQR, 0.00-0.94) events/h. Based on the OAHI, nine patients had no OSA, three had mild OSA, none had moderate OSA, and two had severe OSA. Central respiratory events were observed in nine patients (64.3%); however, all patients had a CAI < 5 events/h, and therefore, none fulfilled the diagnostic criteria for CSAS. The mean oxygen saturation was 92.2 ± 7.3%, with a minimum oxygen saturation of 80.8 ± 12.2%. The median time spent with oxygen saturation below 88% was 8.2 (IQR, 0.3-30.4) min. The mean heart rate was 97.9 ± 20.1 beats/min, whereas the mean minimum and maximum heart rates (available in 11 patients) were 50.3 ± 21.5 and 142.5 ± 20.1 beats/min, respectively. No patient had periodic limb movements during sleep.

Neuroradiological findings were available for 13 patients and are summarized in Table 3. One patient did not undergo MRI because parental consent was not obtained. The most common abnormalities were corpus callosum thinning or atrophy and FM stenosis (7/13, 53.8% each), followed by ventricular dilatation (4/13, 30.8%) and enlarged cerebrospinal fluid spaces (3/13, 23.1%). Cerebellar tonsillar ectopia was identified in three (23.1%) patients. In contrast, basilar invagination, cervical

TABLE 1
Demographic, anthropometric, and clinical characteristics of children with achondroplasia (n = 14)

Variables	n	%	Mean ± SD
Age (year)			5.5 ± 4.3
Sex			
Male	10	71.4	
Weight (kg)			17.1 ± 13.3
Height (cm)			87.1 ± 23.0
Body mass index (kg/m^2)			20.9 ± 9.2
Head circumference (cm)			51.2 ± 4.0
FGFR3 mutation confirmed	14	100	
Foramen magnum stenosis	7	50.0	
Foramen magnum decompression	5	35.7	
Adenoidectomy	3	21.4	
Positive airway pressure therapy	4	28.6	

SD, standard deviation; FGFR3, fibroblast growth factor receptor-3. Adenoidectomy was performed in three children during the available observation period: two before the first PSG and one after the first polysomnography.

TABLE 2
Polysomnographic findings in children with achondroplasia (n = 14)

Variables	n	%	Mean ± SD	Median	IQR
Sleep architecture					
Total sleep time (min)			393.3 ± 59.6		
Sleep efficiency (%)			90.1 ± 8.6		
Sleep latency (min)			9.3 ± 7.3		
REM latency (min)			212.0 ± 113.8		
N1 sleep (%)			4.0 ± 4.2		
N2 sleep (%)			38.1 ± 10.3		
N3 sleep (%)			46.7 ± 13.6		
REM sleep (%)			11.2 ± 5.4		
Respiratory parameters					
Total AHI (events/h)				1.06	0.57–2.93
OAHI (events/h)				0.64	0.24–1.84
REM-AHI (events/h)				2.24	0.00–7.97
CAI (events/h)				0.32	0.00–0.94
Mean oxygen saturation (%)			92.2 ± 7.3		
Minimum oxygen saturation (%)			80.8 ± 12.2		
Time with SpO_2 < 88% (min)				8.2	0.3–30.4
Cardiac parameters					
Mean heart rate (beats/min)			97.9 ± 20.1		
Other PSG findings					
PLM			0.0 ± 0.0		
OSA severity (based on OAHI)					
Normal (OAHI < 1)	9	64.3			
Mild OSA (1 ≤ OAHI < 5)	3	21.4			
Moderate OSA (5 ≤ OAHI < 10)	0	0			
Severe OSA (≥ 10)	2	14.3			
Central respiratory events					
≥ 1 central apnea event	9	64.3			
CAI ≥ 1 events/h	4	28.6			
CAI ≥ 5 events/h (CSAS)	0	0			

SD, standard deviation; IQR, interquartile range; REM, rapid eye movement; N1, N2, and N3, non-REM sleep stages 1, 2, and 3; AHI, apnea-hypopnea index; OAHI, obstructive apnea-hypopnea index; CAI, central apnea index; SpO_2, peripheral oxygen saturation; PSG, polysomnography; PLM, periodic limb movements; OSA, obstructive sleep apnea; CSAS, central sleep apnea syndrome.

spinal cord myelomalacia, vermian hypoplasia/dysgenesis, periventricular white matter volume loss, and frontotemporal cortical atrophy were each observed in one patient (7.7%). Magnetic resonance imaging findings were normal in five patients (38.5%). Achondroplasia Foramen Magnum Score grades were assessed in children with FM stenosis. Two children had AFMS Grade 1, one had Grade 2, one had Grade 3, and three had Grade 4. Five children had undergone FM decompression: one with preoperative AFMS Grade 1, one with Grade 3, and three with Grade 4. Individual PSG findings, AFMS grades, and treatment histories are summarized in Table 4.

Five children had undergone FM decompression before their first PSG. Preoperative PSG data were unavailable for all five children. On postoperative PSG, four (80.0%) children had a total AHI ≥ 1 event/h. Three (60.0%) had OSA based on OAHI, including two with mild and one with severe OSA. One child had an elevated total AHI predominantly attributable to central events (AHI, 1.88; CAI, 1.76; OAHI, 0.13 events/h),

TABLE 3
Neuroradiological findings in children with achondroplasia (n = 13)

Finding	n	%
Normal MRI	5	38.5
Craniocervical abnormalities		
Foramen magnum stenosis	7	53.8
Basilar invagination	1	7.7
Cerebellar tonsillar ectopia/tonsillar herniation	3	23.1
Cervical spinal cord myelomalacia	1	7.7
Intracranial abnormalities		
Corpus callosum abnormality (thinning/atrophy)	7	53.8
Ventricular dilatation	4	30.8
Enlarged cerebrospinal fluid spaces	3	23.1
Vermian hypoplasia/dysgenesis	1	7.7
Periventricular white matter volume loss	1	7.7
Frontotemporal cortical atrophy	1	7.7

Patients may have more than one neuroradiological abnormality. One patient did not undergo Magnetic resonance imaging because parental consent was not obtained.

without meeting the study criterion for CSAS. Positive airway pressure therapy was initiated in four patients (28.6%) during the available follow-up, including CPAP in three and BPAP in one (one of them had no available MRI).

Among the 13 children with available MRI data, central respiratory events were observed in five of seven children with documented FM stenosis (71.4%) and three of six without FM stenosis (50.0%). The difference was not statistically significant (two-sided Fisher's exact test, $p = 0.592$). In the five children who underwent decompression, MRI findings were preoperative, whereas PSG was performed postoperatively.

DISCUSSION

In the present study, we comprehensively evaluated PSG findings and neuroradiological abnormalities in a cohort of children with genetically confirmed achondroplasia. The principal findings were as follows: *(i)* SRBDs were common but were predominantly mild in severity; *(ii)* central

TABLE 4
Patient-level PSG findings, AFMS grades, and treatment history

Patient	AHI (events/h)	CAI (events/h)	OAHI (events/h)	AFMS grade	FM decompression	Decompression-PSG interval (months)*	Adenoid surgery	PAP therapy
P01	19.80	0.00	19.80	4	Yes	13	After first PSG†	None
P02	0.55	0.00	0.55	0	No	N/A	Before first PSG	None
P03	1.11	0.16	0.95	0	No	N/A	None	None
P04	0.45	0.00	0.45	1	No	N/A	Before first PSG	None
P05	3.51	1.56	1.95	4	Yes	2	None	BPAP
P06	0.00	0.00	0.00	0	No	N/A	None	None
P07	2.76	0.58	2.18	1	Yes	36	None; adenoid hypertrophy documented	CPAP
P08	0.88	0.15	0.73	0	No	N/A	None	None
P09	31.30	1.04	30.26	2	No	N/A	None	CPAP
P10	1.88	1.76	0.13	3	Yes	14	None	None
P11	2.99‡	1.49	1.49	MRI N/A	No	N/A	None	CPAP
P12	1.01	0.63	0.38	0	No	N/A	None	None
P13	0.64	0.48	0.16	4	Yes	42	None	None
P14	0.19	0.00	0.19	0	No	N/A	None	None

PSG, polysomnography; AFMS, Achondroplasia Foramen Magnum Score; AHI, apnea-hypopnea index; CAI, central apnea index; OAHI, obstructive apnea-hypopnea index; FM, foramen magnum; PAP, positive airway pressure; BPAP, bilevel positive airway pressure; CPAP, continuous positive airway pressure; MRI, magnetic resonance imaging; N/A, not applicable or not available, as indicated; *, No predecompression PSG recordings were available for the five operated children. Decompression–PSG intervals are approximate where exact surgical dates were unavailable; †, P01 underwent adenoid surgery on 26 May 2025, after the initial PSG. Follow-up PSG in September 2026 showed total AHI 0.30 events/h. Both PSG recordings were obtained after FM decompression; ‡, Continuous PAP therapy at a pressure of 5 cm H_2O was initiated in July 2026, after the initial PSG. For children who underwent FM decompression, AFMS grades refer to preoperative MRI. AFMS 0 indicates absence of FM stenosis and does not necessarily indicate an otherwise normal MRI. Adenoid surgery is classified relative to the first PSG; PAP therapy reflects treatment recorded during available follow-up.

respiratory events were common but generally occurred with a low central apnea burden and were more frequently observed in children with FM stenosis; *(iii)* the most common neuroradiological findings were corpus callosum abnormalities and FM stenosis, followed by ventricular dilatation, with enlarged cerebrospinal fluid spaces and cerebellar tonsillar ectopia occurring equally often; and *(iv)* a substantial proportion of patients required disease-specific interventions, including FM decompression, adenoidectomy, or PAP therapy. These findings highlight the heterogeneous nature of SRBD in achondroplasia and emphasize the importance of multidisciplinary evaluation.

Sleep-related breathing disorders in achondroplasia arise from a complex interaction between craniofacial abnormalities and neurological factors rather than from a single underlying mechanism.[14,15] Midface hypoplasia, maxillary retrusion, upper airway narrowing, and adenotonsillar hypertrophy predispose patients to obstructive respiratory events, whereas cervicomedullary compression secondary to FM stenosis contributes predominantly to central respiratory abnormalities.[16,17] As these mechanisms frequently coexist, current international guidelines advise lifelong respiratory surveillance, with PSG incorporated into routine follow-up whenever clinically indicated.[15,16] Supporting this approach, Ascencio et al.[9] recently demonstrated that OSA developed early in many children with achondroplasia and that timely PSG evaluation was essential for selecting appropriate therapeutic strategies, particularly in patients with moderate-to-severe disease. Excess weight may also contribute to upper airway obstruction. Two children in our cohort had BMI values above the 95th percentile on achondroplasia-specific charts, although these values alone do not establish obesity. Midface hypoplasia and adenotonsillar hypertrophy are additional possible contributors, but their relationship with OSA could not be assessed systematically from the available records.

Obstructive sleep apnea was identified in 35.7% of patients (5/14), including three children with mild disease and two with severe disease. Although sleep-disordered breathing was common, most children either had no OSA or only mild disease according to the OAHI classification. This differs from several previous series reporting a higher proportion of moderate or severe disease. One possible explanation is differences in referral patterns. Unlike earlier studies, in which PSG was frequently performed in children referred due to significant respiratory symptoms, our patients were systematically evaluated within a multidisciplinary follow-up program regardless of disease severity. Consequently, milder forms of OSA may have been detected before the development of more advanced respiratory impairment. This interpretation is consistent with findings from a large single-center study of patients with achondroplasia, although the study population consisted of selected patients referred for sleep evaluation.[18] Similarly, the higher prevalence of severe OSA reported by Ascencio et al.[9] and Buciek et al.[8] may partly reflect referral bias toward children with more pronounced sleep-related symptoms. Therefore, our findings support the growing emphasis on proactive surveillance rather than symptom-driven referral, as recommended in contemporary international guidelines.[15,16,19] Moreover, because our cohort was evaluated after implementation of these updated recommendations, our results may better reflect current real-world clinical practice.

Central respiratory events were numerically more frequent in children with FM stenosis, although this difference was not statistically significant (p = 0.592). The overall central apnea burden remained low, and none of the patients fulfilled the contemporary pediatric diagnostic criteria for CSAS (CAI ≥ 5 events/h). These findings suggest that craniocervical abnormalities may predispose children to central respiratory events without necessarily resulting in clinically significant CSAS.

Sleep-related breathing disorders in achondroplasia cannot be explained solely by FM stenosis. Central respiratory events observed after decompression may have been reduced compared to the preoperative period; however, this possibility could not be assessed because preoperative PSG data were unavailable. Their presence on postoperative PSG does not establish that decompression was ineffective, nor does it allow us to determine whether they remained unchanged or improved. Given the small sample size and the use of preoperative MRI and postoperative PSG in children who underwent decompression, these findings should be interpreted as descriptive and hypothesis-generating rather than as evidence of a causal relationship. Upper airway obstruction and central respiratory abnormalities may coexist and may respond differently to treatment.

Our findings are consistent with previous reports demonstrating that upper airway

obstruction frequently persists despite neurosurgical intervention. Booth et al.[20] reported that OSA often remains after FM decompression and highlighted the contribution of multilevel upper airway abnormalities to persistent respiratory obstruction. Similarly, current international recommendations emphasize that management should not rely solely on neuroimaging findings but should integrate PSG results, neurological evaluation, and clinical assessment when determining treatment strategies.[6,9,15,16] Taken together, our results further support the concept that the respiratory phenotype of achondroplasia is multifactorial and that MRI findings alone are insufficient to predict the severity or pattern of sleep-disordered breathing. Comprehensive PSG evaluation, therefore, remains essential for individualized clinical decision-making. The clinical course of one child provides an illustrative observation regarding the contribution of upper airway obstruction. Despite prior FM decompression, the initial PSG showed severe OSA (OAHI, 19.8 events/h), whereas follow-up PSG after adenoid surgery showed a total AHI of 0.3 events/h. Although this improvement cannot be attributed solely to surgery due to the interval between assessments and possible effects of growth or other clinical changes, it suggests that adenoid-related obstruction may contribute substantially to OSA in some children with achondroplasia. This observation should be interpreted cautiously given the small cohort and the absence of systematic pre- and postoperative PSG assessments for upper airway surgery.

Mean sleep efficiency was 90.1 ± 8.6%. The mean percentages of N1, N2, N3, and REM sleep were 4.0 ± 4.2%, 38.1 ± 10.3%, 46.7 ± 13.6%, and 11.2 ± 5.4%, respectively. Pediatric normative data demonstrate age-related variation in sleep architecture.[21] Given the absence of a healthy control group or an individual age-matched comparison with normative data, these findings are presented descriptively and should not be interpreted as demonstrating normal sleep architecture. In our patient cohort, respiratory events appeared to occur predominantly during REM sleep, suggesting that upper airway obstruction may become more pronounced during this sleep stage. This observation may be explained by the physiological reduction in upper airway muscle tone during REM sleep, which can exacerbate upper airway collapse in children with achondroplasia.

Several limitations should be acknowledged. The retrospective single-center design and small sample size limit the generalizability of our results. The absence of a healthy control group limits the interpretation of sleep architecture findings. Preoperative PSG data were unavailable for children who underwent FM decompression; therefore, changes in respiratory parameters following decompression could not be assessed. Nocturnal carbon dioxide monitoring was not performed, preventing assessment of sleep-related hypoventilation. Upper airway anatomical findings were not systematically assessed across the cohort, limiting evaluation of their relationship with respiratory abnormalities. The arousal index was not calculated and therefore was not reported. Despite these limitations, our findings add to the limited pediatric PSG data available in achondroplasia.

In conclusion, our findings demonstrate that sleep-disordered breathing in children with achondroplasia is heterogeneous and cannot be explained by a single underlying mechanism. The observation of different PSG phenotypes, even among patients with similar FM pathology, further supports the multifactorial nature of respiratory dysfunction and highlights the need for individualized management strategies. Although central respiratory events frequently accompanied FM stenosis, none of the patients fulfilled contemporary pediatric diagnostic criteria for CSAS. Severe OSA may occur in the absence of central apnea, while respiratory abnormalities may be observed on postoperative PSG. These findings emphasize that PSG results should always be interpreted alongside neuroradiological and clinical findings within a multidisciplinary framework. Early diagnosis, timely intervention, including decompression surgery, upper airway surgery, or PAP therapy when indicated, and longitudinal PSG follow-up may contribute to improved long-term neurological and cardiopulmonary outcomes in this vulnerable patient population.

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

Author Contributions: D.K.G.: Concept, study design, data collection or processing, data analysis and interpretation, literature search, manuscript writing, critical revision of the manuscript; C.G.C.E.: Study design, data interpretation, critical revision of the manuscript; E.M.: Patient recruitment, clinical data collection, data interpretation, critical revision of the manuscript; K.A.K.: Concept, study design, supervision, data interpretation, critical revision of the manuscript.

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REFERENCES

1. Mogayzel PJ Jr, Carroll JL, Loughlin GM, Hurko O, Francomano CA, Marcus CL. Sleep-disordered breathing in children with achondroplasia. J Pediatr 1998;132:667-71. doi: 10.1016/s0022-3476(98)70358-0.
2. Tenconi R, Khirani S, Amaddeo A, Michot C, Baujat G, Couloigner V, et al. Sleep-disordered breathing and its management in children with achondroplasia. Am J Med Genet A 2017;173:868-78. doi: 10.1002/ajmg.a.38130.
3. White KK, Parnell SE, Kifle Y, Blackledge M, Bompadre V. Is there a correlation between sleep disordered breathing and foramen magnum stenosis in children with achondroplasia? Am J Med Genet A 2016;170A:32-41. doi: 10.1002/ajmg.a.37385.
4. Wigg K, Tofts L, Benson S, Porter M. The neuropsychological function of children with achondroplasia. Am J Med Genet A 2016;170:2882-8. doi: 10.1002/ajmg.a.37779.
5. Fauroux B, AlSayed M, Ben-Omran T, Boero S, Boon M, Cormier-Daire V, et al. Management of sleep-disordered breathing in achondroplasia: Guiding principles of the European Achondroplasia Forum. Orphanet J Rare Dis 2025;20:233. doi: 10.1186/s13023-025-03717-0.
6. Kubota T, Adachi M, Kitaoka T, Hasegawa K, Ohata Y, Fujiwara M, et al. Clinical practice guidelines for achondroplasia. Clin Pediatr Endocrinol 2020;29:25-42. doi: 10.1297/cpe.29.25.
7. Yalın OÖ, Yılmaz İA, Sungur MA, Doğu O. Obstructive sleep apnea syndrome, periodic limb movements and related factors. Turk J Neurol 2015; 21:90-4.
8. Buciek LH, Jacobsen JR, Raj S, Adams AM, Vandeleur M, Hove HB, et al. Sleep-disordered breathing in children with achondroplasia assessed by polysomnography: A retrospective chart review. Arch Dis Child 2025;111:43-8. doi: 10.1136/archdischild-2025-328595.
9. Ascencio CG, Wani A, Mitchell RB. Achondroplasia and obstructive sleep apnea: Surgical outcomes and comparison to general population. J Clin Sleep Med 2025;21:883-90. doi: 10.5664/jcsm.11574.
10. Cheung MS, Irving M, Cocca A, Santos R, Shaunak M, Dougherty H, et al. Achondroplasia Foramen Magnum Score: Screening infants for stenosis. Arch Dis Child 2021;106:180-4. doi: 10.1136/archdischild-2020-319625.
11. Troester MM, Quan SF, Berry RB, et al; for the American Academy of Sleep Medicine. The AASM Manual for the Scoring of Sleep and Associated Events: Rules, Terminology and Technical Specifications. Version 3. Darien, IL: American Academy of Sleep Medicine; 2023.
12. Garde AJB, Gibson NA, Samuels MP, Evans HJ. Recent advances in paediatric sleep disordered breathing. Breathe (Sheff) 2022;18:220151. doi: 10.1183/20734735.0151-2022.
13. McLaren AT, Bin-Hasan S, Narang I. Diagnosis, management and pathophysiology of central sleep apnea in children. Paediatr Respir Rev 2019;30:49-57. doi: 10.1016/j.prrv.2018.07.005.
14. Pauli RM. Achondroplasia: A comprehensive clinical review. Orphanet J Rare Dis 2019;14:1. doi: 10.1186/s13023-018-0972-6.
15. Savarirayan R, Ireland P, Irving M, Thompson D, Alves I, Baratela WAR, et al. International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. Nat Rev Endocrinol 2022;18:173-89. doi: 10.1038/s41574-021-00595-x.
16. Irving M, AlSayed M, Arundel P, Baujat G, Ben-Omran T, Boero S, et al. European Achondroplasia Forum guiding principles for the detection and management of foramen magnum stenosis. Orphanet J Rare Dis 2023;18:219. doi: 10.1186/s13023-023-02795-2.
17. Zaffanello M, Cantalupo G, Piacentini G, Gasperi E, Nosetti L, Cavarzere P, et al. Sleep disordered breathing in children with achondroplasia. World J Pediatr 2017;13:8-14. doi: 10.1007/s12519-016-0051-9.
18. Soğukpınar M, Demir GÜ, Utine GE, Gönç EN, Özön ZA, Şimşek-Kiper PÖ. Review of patients with achondroplasia: A single-center's experience with follow-up and associated morbidities. Eur J Pediatr 2024;183:3819-29. doi: 10.1007/s00431-024-05643-y.
19. Hoover-Fong J, Scott CI, Jones MC; Committee on genetics. Health supervision for people with achondroplasia. Pediatrics 2020;145:e20201010. doi: 10.1542/peds.2020-1010.
20. Booth KL, Levy DA, White DR, Meier JD, Pecha PP. Management of obstructive sleep apnea in children with achondroplasia: Outcomes of surgical interventions. Int J Pediatr Otorhinolaryngol 2020;138:110332. doi: 10.1016/j.ijporl.2020.110332.
21. Scholle S, Beyer U, Bernhard M, Eichholz S, Erler T, Graness P, et al. Normative values of polysomnographic parameters in childhood and adolescence: Quantitative sleep parameters. Sleep Med 2011;12:542-9. doi: 10.1016/j.sleep.2010.11.011.