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Case Report

Multidisciplinary Orthodontic and Home Sleep Apnea Testing-Based Assessment of Sleep-Disordered Breathing in a Pediatric Patient with Gorlin–Goltz Syndrome: A Case Report

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Abstract

Background: Gorlin–Goltz syndrome is a rare autosomal dominant condition with characteristic craniofacial and odontogenic anomalies. Orofacial alterations in childhood may precede dermatological findings, highlighting the relevance of early orthodontic and functional evaluation. **Objective**: This case describes a multidisciplinary orthodontic and Home Sleep Apnea Testing (HSAT)-based approach for the assessment of craniofacial morphology and sleep-disordered breathing (SDB) risk in a pediatric patient with Gorlin–Goltz syndrome. **Methods**: A 12-year-old male with a genetically confirmed PTCH1 mutation underwent digital intraoral scanning, orthodontic evaluation, and SDB screening using the Pediatric Sleep Questionnaire (PSQ). Following a positive screening score, HSAT with the Philips Alice NightOne® system was performed under specialist supervision. **Results**: The patient showed recurrent odontogenic cysts, a lateral open bite, and unilateral Class II canine relationship. The PSQ score was 0.579, exceeding the validated cut-off of 0.33 and indicating an elevated SDB risk. HSAT findings were suggestive of mild obstructive sleep apnea based on Respiratory Event Index (REI) values (REI 4.7/h), with an isolated SpO_2 nadir of 77% and a maximum recorded apnea duration of 425 s, warranting cautious specialist interpretation and follow-up assessment. **Conclusions**: Integrating orthodontic assessment, digital documentation, validated screening tools, and objective HSAT-based evaluation may support the early recognition of functional compromise in syndromic pediatric patients. Positive screening results should prompt specialist referral and objective sleep assessment, while attended polysomnography remains indicated when comprehensive sleep architecture evaluation or definitive characterization is required.

Keywords: Gorlin–Goltz syndrome; odontogenic cysts; pediatric orthodontics; sleep-disordered breathing; PTCH1 mutation

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1. Introduction

Gorlin–Goltz syndrome, also known as Nevoid Basal Cell Carcinoma Syndrome (NBCCS), is a rare autosomal dominant genetic disorder with an estimated prevalence ranging from 1:57,000 to 1:256,000, with no sex predilection [1–3]. The condition results most commonly from a germline mutation in the PTCH1 gene, a key regulator of the Hedgehog

signaling pathway involved in cell growth and embryonic development [2]. Phenotypic expression is highly variable and may include multiple early-onset basal cell carcinomas, odontogenic cysts of the jaws, skeletal anomalies such as bifid ribs, calcification of the falx cerebri, palmar and plantar pits, and a predisposition to medulloblastoma—particularly in patients with SUFU-related variants [4].

Orofacial manifestations frequently represent the earliest clinical indicators of the syndrome, often preceding dermatological or oncological findings by several years [1,2,5]. Odontogenic keratocysts occur in up to 90% of affected individuals during childhood or adolescence [5,6] and exhibit locally aggressive, recurrent behavior that may necessitate repeated surgical interventions, with potential consequences on maxillofacial growth, dental eruption, and long-term occlusal stability [7–9]. Beyond structural implications, craniofacial morphology is increasingly recognized as a determinant of upper airway dimensions and function. Several studies have demonstrated associations between specific malocclusions—including transverse maxillary deficiency, lateral open bite, and sagittal skeletal discrepancies—and an elevated risk of sleep-disordered breathing (SDB) in pediatric populations [10–12]. In syndromic patients with altered craniofacial development, this risk may be further amplified by skeletal dysmorphism and neuromuscular factors affecting airway patency during sleep [13].

Several case reports and case series have highlighted the importance of multidisciplinary management in patients with Gorlin–Goltz syndrome. Published experiences have described coordinated involvement of maxillofacial surgeons, orthodontists, pediatric dentists, geneticists, dermatologists, and other specialists to address recurrent odontogenic keratocysts, craniofacial abnormalities, dental development disturbances, and long-term surveillance for syndrome-related manifestations. Early diagnosis and continuous interdisciplinary follow-up have been consistently identified as key factors for optimizing functional and clinical outcomes in affected children [3,5,14].

Despite this established relationship, the potential link between the craniofacial anomalies characteristic of Gorlin–Goltz syndrome and functional outcomes such as SDB has received limited attention in the literature. Most existing reports focus on dermatological, genetic, or surgical aspects of the condition, while airway-related functional outcomes remain underexplored—particularly from an orthodontic and pediatric dental perspective. This represents a clinically relevant gap, given the well-documented impact of pediatric SDB on neurocognitive development, cardiovascular health, behavior, and quality of life [15,16]. Within this context, orthodontists and pediatric dentists are uniquely positioned to contribute to the early detection of SDB risk factors, as they routinely assess craniofacial growth, dental development, and occlusal relationships. The integration of validated screening tools into orthodontic evaluation may enable timely identification and specialist referral, supporting a proactive, multidisciplinary management framework.

To our knowledge, reports integrating orthodontic assessment, digital documentation, validated SDB screening, and objective sleep evaluation in pediatric Gorlin–Goltz syndrome remain limited. The aim of this case report is therefore to describe the orthodontic, craniofacial, and sleep-related findings in a pediatric patient with a genetically confirmed diagnosis of Gorlin–Goltz syndrome, illustrating the clinical utility of integrating digital documentation, PSQ-based screening, and Home Sleep Apnea Testing within a coordinated multidisciplinary approach. This report contributes novel functional insight by linking morphologic findings with objective SDB assessment in a comprehensively documented pediatric case.

2. Materials and Methods

2.1. Patient Information

A 12-year-old male patient with a confirmed diagnosis of Gorlin–Goltz syndrome was referred to the Pediatric Dental Unit of Fondazione Policlinico Universitario Gemelli IRCCS (Rome, Italy) for orthodontic evaluation. Genetic testing had previously identified a pathogenic de novo heterozygous mutation in the PTCH1 gene. The patient was the fifth of five siblings, born to non-consanguineous parents from the same geographical region.

Family history was notable for Mediterranean anemia (mother and maternal grandmother), congenital cataracts (mother, maternal grandfather, and maternal great-grandmother), and bipolar cataract in the second-born sister, surgically corrected at age 16. The patient was born at 37 weeks of gestation via cesarean section following a pregnancy complicated by maternal Mediterranean anemia and thalassemia; neonatal jaundice required phototherapy. At one month of age, he was hospitalized for tracheomalacia. His medical history was significant for multiple maxillofacial surgical interventions related to recurrent odontogenic keratocysts, with no additional systemic comorbidities beyond those associated with Gorlin–Goltz syndrome. The patient was followed at our institution from October 2025 to April 2026, corresponding to an overall follow-up period of approximately six months. Written informed consent was obtained from the patient's legal guardians prior to clinical examination and data collection, in accordance with the Declaration of Helsinki. This case report was prepared following the CARE (CAse REport) guidelines [17].

2.2. Orthodontic and Digital Assessment

Following informed consent, the patient underwent a comprehensive clinical and orthodontic evaluation. Extraoral examination assessed facial symmetry, profile type, and midline relationships. Intraoral examination focused on dental eruption status, the presence of missing or impacted teeth, occlusal relationships, and transverse and vertical discrepancies. Malocclusion was classified according to Angle's criteria [7]. Maxillary transverse evaluation included assessment of arch form and symmetry, palatal morphology, breathing pattern (nasal versus oral), and buccal corridor width while smiling. Vertical relationships were evaluated through measurement of incisal overbite. Digital orthodontic documentation was obtained using the iTero® intraoral scanner (Align Technology, San Jose, CA, USA). Digital scans provided high-resolution, three-dimensional models of the dental arches, enabling detailed morphological assessment of tooth position, arch dimensions, interocclusal relationships, and space conditions. Standardized extraoral and intraoral photographic records complemented the digital workflow to ensure comprehensive baseline documentation.

2.3. Sleep-Disordered Breathing Screening

Risk of sleep-disordered breathing was assessed using the validated Italian version of the Pediatric Sleep Questionnaire (PSQ) (Figure 1), a parent-report instrument with established sensitivity and specificity for identifying SDB risk in pediatric populations [15,18]. The questionnaire was completed by the patient's caregivers during the orthodontic consultation. The PSQ score was calculated as the proportion of affirmative responses relative to the total number of answered items, following the original scoring method described by Chervin et al. [16]. Unanswered items were excluded from the denominator. A total score exceeding 0.33 was considered indicative of elevated SDB risk, warranting further diagnostic evaluation. It should be noted that the PSQ is a validated screening tool rather than a diagnostic instrument; therefore, results were interpreted exclusively as indicators of SDB risk requiring objective confirmation.

Pediatric Sleep Questionnaire
(Screening)

Name of the child:_____ Date of birth:_____
Person completing this form: _____
Date that you are completing the questionnaire: _____

Instructions: Please answer the questions about how your child **IN THE PAST MONTH**. Circle the correct response or *print* your answers in the space provided. "Y" means "yes," "N" means "no," and "DK" means "don't know." For this questionnaire, the word "usually" means "more than half the time" or "on more than half the nights."

Please answer the following questions as they pertain to your child in the past month.

		YES	NO	Don't Know
1.	**While sleeping, does your child:**			
	Snore more than half the time?	Y	N	DK
	Always snore?	Y	N	DK
	Snore loudly?	Y	N	DK
	Have "heavy" or loud breathing?	Y	N	DK
	Have trouble breathing, or struggle to breath?	Y	N	DK
2.	**Have you ever seen your child stop breathing during the night?**	Y	N	DK
3.	**Does your child:**			
	Tend to breathe through the mouth during the day?	Y	N	DK
	Have a dry mouth on waking up in the morning?	Y	N	DK
	Occasionally wet the bed?	Y	N	DK
4.	**Does your child:**			
	Wake up feeling unrefreshed in the morning?	Y	N	DK
	Have a problem with sleepiness during the day?	Y	N	DK
5.	**Has a teacher or other supervisor commented that your child appears sleepy during the day?**	Y	N	DK
6.	**Is it hard to wake your child up in the morning?**	Y	N	DK
7.	**Does your child wake up with headaches in the morning?**	Y	N	DK
8.	**Did your child stop growing at a normal rate at any time since birth?**	Y	N	DK
9.	**Is your child overweight?**	Y	N	DK
10.	**This child often:**			
	Does not seem to listen when spoken to directly	Y	N	DK
	Has difficulty organizing tasks and activities	Y	N	DK
	Is easily distracted by extraneous stimuli	Y	N	DK
	Fidgets with hands or feet, or squirms in seat	Y	N	DK
	Is "on the go" or often acts as if "driven by a motor"	Y	N	DK
	Interrupts or intrudes on others (eg butts into conversations or games)	Y	N	DK

Figure 1. Pediatric Sleep Questionnaire (PSQ), a validated screening tool for identifying children at risk of sleep-disordered breathing.

2.4. Objective Sleep Assessment

Following a positive PSQ screening result, the patient was referred for specialist evaluation and objective sleep assessment. Home Sleep Apnea Testing (HSAT) was performed using the Philips Alice NightOne® device (Philips Respironics, Amsterdam, The Netherlands), a level III portable monitoring system used for ambulatory respiratory sleep assessment under specialist supervision. The device recorded the following parameters throughout the night: respiratory effort through thoracic and abdominal movement, nasal airflow via a pressure transducer, pulse oximetry (SpO_2), body position, and snoring intensity. Total recording time was documented, while true total sleep time could not be determined because electroencephalographic sleep staging was not available. Respiratory events were classified according to the pediatric scoring rules of the American Academy of Sleep Medicine Manual for the Scoring of Sleep and Associated Events [17]. The HSAT recording, including airflow, respiratory effort, oximetry signals, and automatically detected respiratory events, was reviewed for signal quality and artifact exclusion before final interpretation. The tracing was formally reviewed by a sleep medicine specialist before final clinical interpretation. Particular attention was given to prolonged events, oxygen desaturation nadir, and the classification of obstructive versus central events. The Respiratory Event Index (REI) was calculated as the total number of apneas and hypopneas divided by the total recording time in hours. Because total sleep time could not be determined, REI was based on recording time rather than actual sleep time; therefore, the severity of sleep-disordered breathing may have been underestimated or overestimated. In pediatric patients, an REI $\geq$ 1 event/hour is considered abnormal, with severity graded as mild (1–5/h), moderate (5–10/h), or severe (>10/h). The Oxygen Desaturation Index (ODI) was defined as the number of $\geq$3% oxygen desaturation events per hour of recording. Additional parameters analyzed included mean and nadir SpO_2, maximum apnea duration, and positional distribution of respiratory events. The selection of HSAT over attended polysomnography (PSG) was based on clinical accessibility, patient compliance considerations, and the utility

of level III monitoring for initial SDB characterization in pediatric patients with a high pre-test probability following positive screening [17,19]. Limitations inherent to HSAT methodology—including the absence of electroencephalographic sleep staging and the use of recording time rather than true total sleep time for REI calculation—were acknowledged, and follow-up PSG was recommended for comprehensive sleep architecture evaluation.

3. Case Presentation and Clinical Findings

3.1. Medical History and Surgical Intervention

Between 2023 and 2025, the patient underwent multiple maxillofacial surgical procedures for the removal of recurrent odontogenic keratocysts affecting both the maxilla and mandible. These interventions resulted in the extraction of eight impacted teeth and tooth germs across both arches. Several lesions involved or communicated with the maxillary sinus, requiring coordinated surgical planning. Table 1 summarizes the timing and location of surgical extractions performed during this period. Cone-beam computed tomography (CBCT) imaging obtained in 2025, including panoramic reconstruction views, revealed multiple residual and recurrent cystic lesions, with prominent involvement of the first and third quadrants affecting teeth 1.5, 1.6, 1.7, and 3.5 (Figure 2).

Table 1. Chronological summary of maxillofacial surgical interventions and associated extractions in the patient with Gorlin–Goltz syndrome. * = Tooth germs.

Year	Extractions of Impacted Teeth
2023	2.3
2024	4.4
2025	1.5, 1.6, 1.8 *; 2.8 *, 3.5, 4.8 *

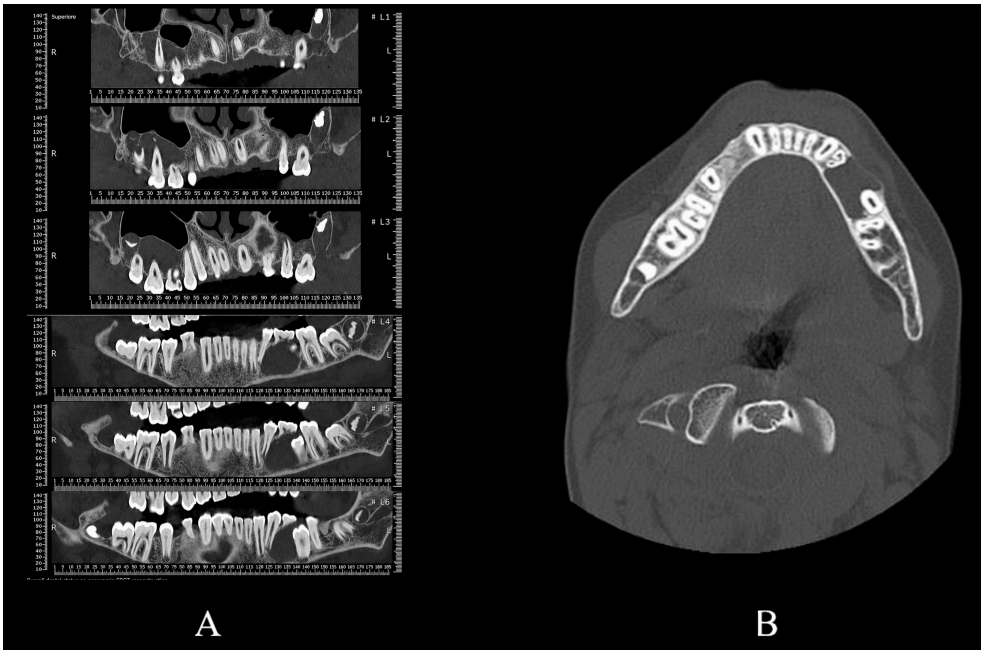

Figure 2. CBCT imaging. (**A**) Panoramic reconstruction showing the patient's dental status and recurrent odontogenic cysts. (**B**) Axial CBCT section showing the largest mandibular odontogenic cystic lesion.

3.2. Clinical and Orthodontic Findings

Extraoral examination revealed an orthognathic skeletal profile with adequate facial proportions. Facial and mandibular midlines were coincident; however, the maxillary

dental midline was deviated to the left. No significant facial asymmetry was observed. Intraoral examination documented multiple missing teeth secondary to surgical extraction or agenesis. Occlusal analysis demonstrated an Angle Class II canine relationship on the right side and a lateral open bite on the left, with absence of posterior occlusal contact in the second quadrant. Overbite and overjet were within normal limits in the anterior region. Standardized extraoral and intraoral photographic records documented these findings (Figure 3). Digital intraoral scanning using the iTero® system provided high-resolution three-dimensional models of both dental arches, enabling precise assessment of tooth position, arch form, interarch relationships, and residual spacing. The digital models confirmed maxillary arch asymmetry with a reduced transverse dimension in the left posterior segment and irregular spacing distribution secondary to prior extractions (Figure 4).

Figure 3. (**A**) Extra-oral photographic records of the patient. (**B**) Intra-oral photographic records of the patient.

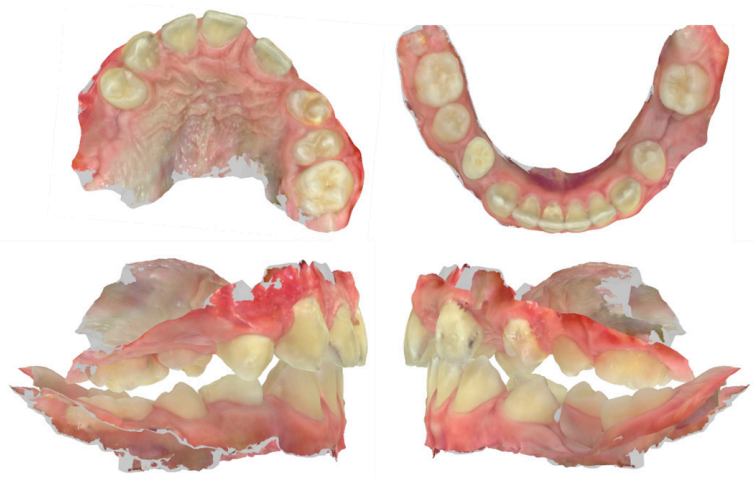

Figure 4. Digital intraoral scan acquired using the iTero® system (Align Technology, San Jose, CA, USA).

3.3. Sleep-Disordered Breathing Questionnaire

The Pediatric Sleep Questionnaire (PSQ), completed by the patient's caregivers yielded a total score of 0.579, substantially exceeding the validated cut-off threshold of 0.33. All three PSQ subscales were positive: Snoring, Sleepiness/Daytime Somnolence, and

Behavior/Inattention-Hyperactivity. This result indicated an elevated screening-positive risk of sleep-disordered breathing, prompting referral for specialist evaluation and objective sleep assessment.

3.4. Objective Sleep Assessment Outcomes

Home Sleep Apnea Testing (HSAT) was performed using the Philips Alice NightOne® device over a single night with adequate recording quality. The total recording time was 7.2 h. The Respiratory Event Index (REI) was 4.7 events per hour, an HSAT-based finding consistent with mild obstructive sleep apnea according to AASM pediatric criteria. A total of 22 respiratory events were recorded. The HSAT-derived indices included an Obstructive Apnea Index of 1.9/h and a Central Apnea Index of 1.9/h. The central component was reported as an HSAT-derived parameter and interpreted cautiously, without establishing a diagnosis of central sleep apnea. Key respiratory and oxygenation parameters are summarized in Table 2.

Table 2. Summary of Home Sleep Apnea Testing results.

Parameter	Value
Total recording time	7.2 h
Respiratory Event Index (REI)	4.7/h
Obstructive Apnea Index (OAI)	1.9/h
Central Apnea Index (CAI)	1.9/h
Mean apnea duration	61.3 s
Maximum apnea duration	425 s
Mean SpO_2	98%
Nadir SpO_2	77%
Oxygen Desaturation Index (ODI)	2.9/h
Snoring (% total time)	0%

Although the mean oxygen saturation remained stable at 98%, the HSAT recording showed an isolated SpO_2 nadir of 77% in association with a prolonged respiratory event. Given the ambulatory nature of the recording, this finding was interpreted after review of signal quality and artifact exclusion and should be considered clinically relevant but requiring confirmation in the context of specialist sleep evaluation. The respiratory disorder exhibited a strongly positional profile. Of the 22 recorded events, 19 (86%) occurred in the supine position, corresponding to a supine REI of 5.4/h compared with 0.9/h in the left lateral decubitus position (Table 3).

Table 3. Positional distribution of respiratory events.

Body Position	Events (n)	REI (/h)
Supine	19	5.4
Left lateral	3	0.9
Right lateral	0	-
Prone	0	-

The complete absence of snoring throughout the recording (0% of total time) was a notable finding, demonstrating that clinically significant upper airway obstruction may occur without audible respiratory sounds in pediatric patients.

4. Discussion

This case report illustrates the complex phenotypic expression of Gorlin–Goltz syndrome in a pediatric patient, with particular emphasis on the interplay between recurrent

odontogenic lesions, craniofacial morphology, occlusal development, and functional implications related to upper airway patency. The principal contribution of this report lies in the documentation of a structured pathway from validated screening to objective ambulatory sleep assessment: a validated questionnaire identified an elevated risk of sleep-disordered breathing, prompting specialist referral and HSAT-based respiratory evaluation, which showed findings consistent with mild OSA-compatible respiratory disturbance. This case illustrates the feasibility of integrating orthodontic assessment, digital documentation, validated SDB screening, and objective ambulatory sleep evaluation within a multidisciplinary framework. Gorlin–Goltz syndrome is characterized by marked phenotypic variability, even among patients carrying identical pathogenic mutations [4,18]. This heterogeneity complicates early diagnosis and longitudinal management, particularly in pediatric patients in whom craniofacial and dental manifestations frequently precede cutaneous or oncological signs by several years [1,2]. Odontogenic keratocysts, reported in up to 90% of affected individuals during childhood or adolescence [5,6], exhibit locally aggressive and recurrent behavior that often necessitates repeated surgical interventions. The cumulative impact of these procedures on maxillofacial growth, dental eruption, and occlusal stability underscores the importance of continuous orthodontic monitoring throughout development [7–9].

Beyond structural and occlusal implications, craniofacial morphology is increasingly recognized as a determinant of upper airway dimensions and function. Multiple studies have demonstrated associations between specific malocclusions—including lateral open bite, transverse maxillary deficiency, and sagittal skeletal discrepancies—and elevated SDB risk in pediatric populations [10,11]. In syndromic patients with altered craniofacial development, this risk may be further amplified by skeletal dysmorphism and neuromuscular factors affecting airway stability during sleep [12].

Published evidence specifically addressing sleep-disordered breathing in patients with Gorlin–Goltz syndrome remains extremely limited. Most available reports focus on odontogenic keratocysts, skeletal abnormalities, dermatological manifestations, and genetic aspects of the syndrome, whereas functional airway outcomes have received comparatively little attention. Although craniofacial characteristics frequently observed in Gorlin–Goltz syndrome—including maxillary constriction, dental anomalies, altered occlusal relationships, and disturbances of craniofacial growth—have been associated with an increased risk of upper airway dysfunction in the general pediatric population, objective sleep assessments have rarely been reported in affected individuals. Consequently, direct comparison with previously published cases is limited and highlights the need for further studies investigating airway function and sleep-related outcomes in this rare disorder. Home Sleep Apnea Testing provided objective ambulatory respiratory data that strengthened the functional interpretation of this case beyond descriptive craniofacial documentation. The HSAT-derived REI of 4.7 events per hour supported a finding of mild OSA according to pediatric criteria. However, aggregate indices alone do not capture the full clinical picture.

Event-level parameters required careful interpretation because the HSAT showed a prolonged respiratory event and an isolated SpO_2 nadir of 77% despite a mean oxygen saturation of 98% and an overall REI in the mild range. These findings highlight that summary indices may not fully capture the clinical relevance of individual events. However, because they were obtained through ambulatory level III monitoring, they should be interpreted in the context of signal-quality review, artifact exclusion, and specialist sleep assessment. Follow-up attended polysomnography would be useful to confirm event severity, further characterize respiratory-event type, and evaluate sleep architecture [19].

The strong positional nature of the respiratory disorder represents an actionable finding. With 86% of events occurring in the supine position (supine REI 5.4/h versus 0.9/h in

the lateral decubitus), positional therapy emerges as a low-burden initial management strategy. This approach may serve as a temporizing measure while multidisciplinary planning addresses longer-term orthodontic and surgical considerations. The complete absence of snoring during the recording warrants specific comment. This finding demonstrates that in pediatric patients—particularly those with syndromic craniofacial features—significant upper airway obstruction may occur silently, without the auditory cues that typically prompt parental concern and clinical referral. This observation reinforces the value of systematic screening protocols that do not rely solely on symptom-driven identification.

Because the assessment was performed using a level III HSAT device, electroencephalographic monitoring was not available. Consequently, sleep-stage distribution, REM-related respiratory events, arousal indices, sleep efficiency, and overall sleep architecture could not be evaluated. These limitations should be considered when interpreting the reported respiratory findings. For this reason, attended polysomnography remains indicated if comprehensive sleep architecture assessment, REM-related event characterization, or confirmation under controlled laboratory conditions is required.

In this case, digital intraoral scanning and standardized photographic documentation served as supportive tools for reproducible baseline recording and multidisciplinary communication. Three-dimensional digital models facilitated the visualization of arch asymmetry, spacing irregularities, and occlusal discrepancies, complementing the clinical examination rather than replacing specialist diagnostic assessment.

From a practical standpoint, the ability to share objective craniofacial data with sleep medicine specialists supports more informed interpretation of functional risk factors. Digital records also establish a reliable baseline for longitudinal monitoring of craniofacial growth and treatment response. While the specific contribution of digital workflow to clinical outcomes requires systematic evaluation in larger cohorts, this case illustrates its potential utility within an integrated assessment protocol.

This case supports several clinically relevant recommendations. Although conclusions cannot be drawn from a single case, this observation suggests that clinicians managing pediatric patients with Gorlin–Goltz syndrome may consider incorporating validated SDB screening tools when craniofacial risk factors or suggestive symptoms are present. Positive screening findings should prompt referral for specialist evaluation and consideration of objective sleep assessment according to clinical judgment. Home sleep apnea testing (HSAT) represents an accessible initial option, although attended polysomnography remains the gold standard for a comprehensive evaluation. In patients with strongly positional obstructive sleep apnea, positional therapy may provide symptomatic relief while definitive management is being planned.

In the present case, multidisciplinary management involved pediatric dentistry, orthodontics, maxillofacial surgery, clinical genetics, and sleep medicine specialists. Sleep medicine consultation guided the indication for HSAT and interpretation of respiratory findings, whereas maxillofacial surgeons monitored recurrent odontogenic cysts and associated surgical needs. Although additional specialties such as otolaryngology and pulmonology were not directly involved at the time of assessment, their contribution may be valuable in future airway-oriented evaluations. This report carries inherent limitations as a single-patient observation. Findings cannot be generalized to the broader Gorlin–Goltz population, and individual variability in phenotypic expression limits extrapolation. The HSAT methodology, while useful for ambulatory respiratory sleep assessment, does not replace attended polysomnography as the gold standard for comprehensive sleep staging and full diagnostic characterization. Additionally, the single-night recording may not capture night-to-night variability in respiratory parameters. Additional limitations should also be acknowledged. Information regarding adenotonsillar status was not systematically

collected and therefore could not be included in the present report, despite its recognized relevance in the pathophysiology of pediatric obstructive sleep apnea. Furthermore, a formal otolaryngological evaluation was not available, preventing comprehensive assessment of potential upper airway anatomical contributors to sleep-disordered breathing. Objective airway investigations, such as nasoendoscopy, rhinomanometry, acoustic pharyngometry, or other dedicated upper airway assessments, were likewise not performed. Consequently, the anatomical site and relative contribution of specific upper airway obstruction mechanisms could not be directly characterized. Future multidisciplinary studies incorporating ENT examination and objective airway assessment may provide a more comprehensive understanding of sleep-disordered breathing risk in patients with Gorlin–Goltz syndrome. Nonetheless, the use of standardized clinical documentation, a validated screening instrument with established psychometric properties, and objective ambulatory monitoring strengthens the reproducibility and clinical relevance of the observations presented.

Longitudinal and multicenter studies are needed to define the true prevalence of SDB in Gorlin–Goltz syndrome and to identify phenotypic or genotypic predictors of airway compromise. Prospective evaluation of integrated screening protocols—combining digital orthodontic assessment with validated SDB questionnaires—may determine whether such approaches meaningfully improve functional outcomes and reduce diagnostic delay in affected children. Additionally, investigation of the relationship between specific craniofacial parameters (quantified through digital cephalometric and three-dimensional analysis) and objective sleep metrics could refine risk stratification and support targeted early intervention. Registry-based approaches may facilitate the accumulation of sufficient case numbers to address these questions in a rare disease population.

5. Conclusions

Integrating orthodontic assessment, digital documentation, validated screening tools, and objective HSAT-based evaluation may support the early recognition of functional compromise in syndromic pediatric patients. Positive screening results should prompt specialist referral and objective sleep assessment, while attended polysomnography remains indicated when comprehensive sleep architecture evaluation or definitive characterization is required.

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