

Sleep-Disordered Breathing in Children with Rare Skeletal Disorders: A Survey of Clinical Records

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Significance of the Study

- Sleep-disordered breathing has been confirmed in children with achondroplasia, and adenotonsillectomy could not revert such condition. In a subgroup of patients with achondroplasia, the respiratory disturbance has been correlated with anatomical characteristics of the upper airways. Nevertheless, in children with *Osteogenesis Imperfecta* and Ellis van Creveld Syndrome, the respiratory disturbance during sleep was mild but often present.

Keywords

Sleep-disordered breathing · Obstructive sleep apnea · Achondroplasia · Osteogenesis imperfecta · Ellis van Creveld syndrome · Overnight respiratory polygraph

Abstract

Objective: Craniofacial disharmony in skeletal diseases is strongly associated with sleep-disordered breathing. This study was aimed at studying the sleep respiratory patterns in young children with rare skeletal disorders. **Design:** This retrospective study included children with achondroplasia (ACH), osteogenesis imperfecta (OI) and Ellis van Creveld Syndrome. Our subjects underwent an in-laboratory over-

night respiratory polygraph between January 2012 and April 2016. All medical records were reviewed and brain Magnetic Resonance Imaging was conducted on patients with ACH, nasopharynx, oropharynx and laryngopharynx spaces. **Patients:** Twenty-four children were enrolled, 13 with ACH, 2 with spondyloepiphyseal dysplasia, 1 with odontochondrodysplasia, 6 with OI and 2 with Ellis van Creveld Syndrome. **Results:** Children with ACH, who had adenotonsillectomy, showed fewer sleep respiratory involvement than untreated children. Among 13 patients with ACH, brain magnetic resonance imaging was available in 10 subjects and significant negative correlation was found between sleep respiratory patterns, nasopharynx and oropharynx space ($p < 0.05$). In 2 patients with spondyloepiphyseal dysplasia, mild-to-mod-

erate sleep respiratory involvement was found. Both subjects had a history of adenotonsillectomy. Mild sleep respiratory involvement was also observed in 4 out of 6 patients with OI. One patient with Ellis van Creveld syndrome had mild sleep respiratory disturbance. **Conclusions:** Sleep respiratory disturbances were detected in children with ACH, and with less severity also in OI and Ellis van Creveld syndrome. Adenotonsillectomy was successful in ACH in reducing symptoms. In light of our findings, multicenter studies are needed to obtain further information on these rare skeletal diseases.

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Published by S. Karger AG, Basel

Introduction

Obstructive sleep apnea (OSA) is characterized by partial or complete obstruction of airflow and oxygen desaturation during sleep owing to upper airway collapse [1]. In children, sleep-disordered breathing (SDB) leads to sleep-related daytime and nighttime symptoms [2]. The severity of sleep apnea depends on the size of the high airway, and neuromuscular control also plays a role [3]. In children, the major contributor to airway narrowing is hyperplasia of pharyngeal tonsils and adenoids, and pediatric craniofacial disharmony is strongly associated with SDB [4].

It is known that skeletal dysplasia, such as achondroplasia (ACH; MIM: 100800), is characterized by an increased risk of SDB [5–7]. In particular, the most important breathing disorder in children with ACH is upper airway obstruction during sleep [8]. Subjects with ACH present rhizomelia, macrocephaly, and facial hypoplasia, dysplasia of the skull base and foramen magnum stenosis with cervical spinal cord compression [9], pectus excavatum, thoracic kyphosis, and lumbar lordosis [5]. Enlarged tonsils and adenoids often play a significant role in worsening the upper airway obstruction [10]. Persisting significant OSA after adenotonsillectomy (A&T) [11] is related to the reduced base of the skull and hypoplasia of the middle third of the face in subjects with ACH [12]. Guidelines recommended an early detection of sleep disorders in children with ACH [13].

To our knowledge, the association between skeletal diseases, such as osteogenesis imperfecta (OI) and Ellis-van Creveld syndrome (EVC), and SDB has not been studied in children. OI (MIM: OI III 259420; OI IV 166220; OI VI 613982) is the most commonly inherited connective tissue disorder affecting the bone [14], and the major clinical expression is skeletal fragility [15]. How-

ever, dentinogenesis imperfecta and cranial malformations such as macrocephaly, hydrocephalus, and basilar invagination have been described in subjects with OI [16]. Besides, EVC (MIM 225500) is a chondral and ectodermal dysplasia characterized by short ribs, polydactyly, growth retardation, and heart deficiencies. In EVC, there are also wide oral expressions, including malocclusion [17].

It is not clear from the literature whether both facial and skull differences in OI and EVC are associated with SDB, as well as with ACH; the aim of our study was to conduct the sleep respiratory investigations on these rare skeletal disorders in the clinical setting.

Subjects and Methods

Study Population

Our retrospective study included subjects younger than 15 years old with skeletal diseases (ACH, OI, EVC) who attended the in-laboratory overnight polygraph study between January 2012 and April 2016, admitted for assessment, treatment, and clinical follow-up at the Verona Center for Skeletal Dysplasia (North-East Italy). Twenty-four children were included. Thirteen children with ACH, 2 children with spondyloepiphyseal dysplasia, 1 child with odontochondrodysplasia, 2 children with OI type III, 1 child with OI type IV, 3 children with OI type VI, and 2 children with EVC. All the children with ACH carried a mutation in the FGFR3 gene. Based on interview with the parents, all the children except one with odontochondrodysplasia, had sleep respiratory problems. Respiratory symptoms were detected using the Pediatric Sleep Evaluation Questionnaire [18] when the children came for the polygraph study. The diagnosis was proved based on both clinical characteristics of ACH, OI, EVC, and genetic analysis (i.e., ACH with FGFR3 gene sequencing and OI with type I collagen gene sequencing). The pharmacological treatment for OI subjects was intravenous bisphosphonate [19].

In our subjects with skeletal dysplasia, we analyzed the clinical characteristics of diseases, and studied the course and result of diseases over a follow-up period [20]. The study was performed in agreement with the Declaration of Helsinki and under the terms of all relevant local legislation.

In-Laboratory Overnight Respiratory Polygraph

The in-laboratory overnight respiratory polygraph is reported to be a useful and reliable approach for the diagnosis of OSA syndrome in children [21].

Our subjects underwent an in-laboratory overnight respiratory polygraph study (SOMNOscreen™ PSG, SOMNOmedics GmbH, Randersacker, Germany). The polygraph study recorded nasal airflow, chest and abdominal respiratory movements (thoracic and abdominal belts), arterial oxygen saturation (SaO₂; digital pulse oximetry), heart rate (HR; finger probe), ECG, body position (mercury sensor), and tracheal sounds (microphone). As an index of inspiratory effort, thoracoabdominal asynchrony (phase angle), a vector of rib cage and abdominal respiratory movements, was recorded during natural nocturnal active and quiet sleep. The tho-

racoabdominal asynchrony analysis determined the degree of obstruction calculated from 2 effort signals, thoracic and abdominal belts; when the value is higher, it indicates an increasing degree of inspiratory effort. The device was applied between 6:00 p.m. and 8:00 a.m. of the following day and the data recorded continuously. All children underwent recordings for ≥ 6 h in a quiet and properly prepared sleep room in the presence of one of their parents. Analysis of the entire recording session was performed by visual inspection (DOMINO[®] software, Somnomedics version 2.6.0) [22]. Scoring was performed according to previously reported roles [23, 24].

Baseline was considered T0 – the time at which each child performed the first sleep respiratory test. If children performed more than one sleep respiratory study, the last follow-up measurement was defined as T1.

Magnetic Resonance Imaging

The MRIs reviewed were carried out using 1.5T MR equipment (Siemens Magnetom Symphony, Siemens, Erlangen, Germany). In particular, standard examination included a T1-weighted 3D MP-RAGE sequence (TR = 1,790 ms, TE = 4 ms, matrix = 512 × 512, FA = 15°, Voxel = 1 × 1 × 1 mm), covering the entire brain to the upper part of the neck. Airway volumetric evaluation was done with the semi-automatic growing region segmentation method included in the ITK-SNAP freeware software version 3.4.0 (<http://www.itksnap.org/>) [25]. The air columns of the airways in this sequence were markedly hypointense, showing a strong contrast compared to the nearby anatomical structures. In particular, after loading the software for the T1-weighted 3D MP-RAGE sequence, 3 anatomical regions were selected: the nasopharynx (from the choanae to the upper edge or soft palate), the oropharynx (from the soft palate to the upper edge of the epiglottis), and the laryngopharynx (from the upper edge of the epiglottis to the lower edge of the cricoid cartilage), respectively. A seed was placed in each region and allowed to grow inside the airways. Because of the high contrast between airways and close structures, the segmentation did not grow outside the air columns. At the end, the segmentation was inspected and the volume of each of the 3 regions estimated in mm³.

Statistical Analysis

Statistical analysis was performed using SPSS Statistics 22.0[®] software for Windows. Descriptive statistics (mean, SD, and range) were performed for the quantitative variables considered in this study. The Mann-Whitney test was used between selected subgroups to explore differences between continuous measures. The strength of the association between 2 quantitative variables was evaluated by calculating the simple correlation coefficient (Pearson's test). The value of statistical significance was set at $p < 0.05$.

Results

Study Population

At T0, 13 of the children with ACH had brain MRI performed on them and 11 out of 13 had undergone SEP. The 2 subjects with spondyloepiphyseal dysplasia had brain MRI and one of these had SEP; 1 of 6 children with

OI had brain MRI. None of the 2 EVC children underwent brain MRI and SEPs.

At T0, 3 out of 13 children with ACH (1 male) aged 4.3, 13.4, and 14.6 years, had adenotonsillectomy (at 3.3, 9.0, and 4.0 years old, respectively). However, both children with spondyloepiphyseal dysplasia (5.2 and 9.6 years old) had adenotonsillectomy (at 2 and 4.5 years old, respectively). None of the children with other skeletal diseases (odontocondrodysplasia, OI, and EVC) had undergone adenotonsillectomy. All the children with OI received intravenous bisphosphonate therapy; 3/6 children with OI (all with type VI) had surgery for skeletal fractures.

The angle of obstruction in the supine position was compared between ACH and OI patients at T0. In particular, data were available for 10 out of 24 ACH and 5 out of 6 OI subjects. In ACH patients, the mean angle in the supine position was 24.7 ± 7.7 degrees (supine time $43.2 \pm 28.5\%$) and in OI it was 40.6 ± 26.1 degrees (supine time $78.1 \pm 20.4\%$).

After enrollment, 4 additional children with ACH were referred for adenotonsillectomy; one ACH patient (5 years old) and one ACH male (5.5 years old) were treated with nasal C-PAP and rapid maxillary expansion for persistent OSAS. All the ACH children ($n = 7$) with persistent mild OSAS, not selected for adenotonsillectomy, were treated using nasal steroids. None of the children with ACH underwent neurosurgery for foramen magnum restriction before and during the T0–T1 periods, based on negative results of brain MRI and SEP.

In-Laboratory Overnight Respiratory Polygraph

During the 4-year observational period, more than one polygraph study was performed in 7 out of 13 of the children with ACH and in 2 children with spondyloepiphyseal dysplasia and odontocondrodysplasia. All the children with OI and ECV had one polygraph study. The respiratory features of 3 ACH children who had adenotonsillectomy before evaluation ranged as apnea-hypopnea index (AHI) 1.5–3.7 events/h and oxygen desaturation index (ODI) 1.1–2.6 events/h, considered as follow-up measures after surgery. Four ACH children (3 males) 3.3, 3.2, 3.6, and 2.6 years, respectively, with moderate-severe AHI (8.3, 20.1, 27.3, and 20.9 events/h) were immediately referred for adenotonsillectomy. One child (male, 2.6 years) was not available for follow-up surgery because he was referred to another center. Figure 1 shows the plotted distribution of the AHI (events/h) and ODI (events/h) values, respectively, for the population studied, related to age evaluation.

Table 1. Results of polygraph study in children with achondroplasia. Before enrolment, 3 children (1 male; age 4.30, 13.4, and 14.6 years) had had an A&T (at 3.3, 9, and 4 years, respectively). and the respiratory parameters before the polygraph measures (AHI 1.5–3.7; ODI 1.1–2.6 events/h) were included as follow-up measures (after adenotonsillectomy)

Timing	Achondroplasia (no A&T) Baseline, mean ± SD	Achondroplasia (no A&T) Follow-up, mean ± SD	Achondroplasia (after A&T) Follow-up, mean ± SD
Patients	10	7	6
Age, years	4.8±4.1	7.0±5.1	8.6±4.1
Age at surgery, years	–	–	4.7±2.2
eTST, h	9.22±1.5	8.97±1.2	9.0±1.0
OA, events/h	2.7±3.4	0.35±0.4	0.8±1.0
CA, events/h	1.1±1.5	1.6±1.6	0.5±0.6
H, events/h	5.6±7.2	1.0±1.0	0.6±0.4
AHI, events/h	9.5±9.6	3.2±1.9	2.1±1.1
ODI, events/h	13.7±21.2	2.5±2.0	1.6±0.8
Mean SpO ₂ , %	94.9±4.4	97.7±0.6	95.8±1.6
Min SpO ₂ , %	75.2±18.3	89.3±2.5	88.8±2.9
Snoring (% eTST)	3.1±4.6	3.1±4.7	0.9±1.8

AHI, apnea-hypopnea index; H, hypopnea; OA, obstructive apnea; CA, central apnea; ODI, oxygen desaturation index; eTST, estimated total sleep time.

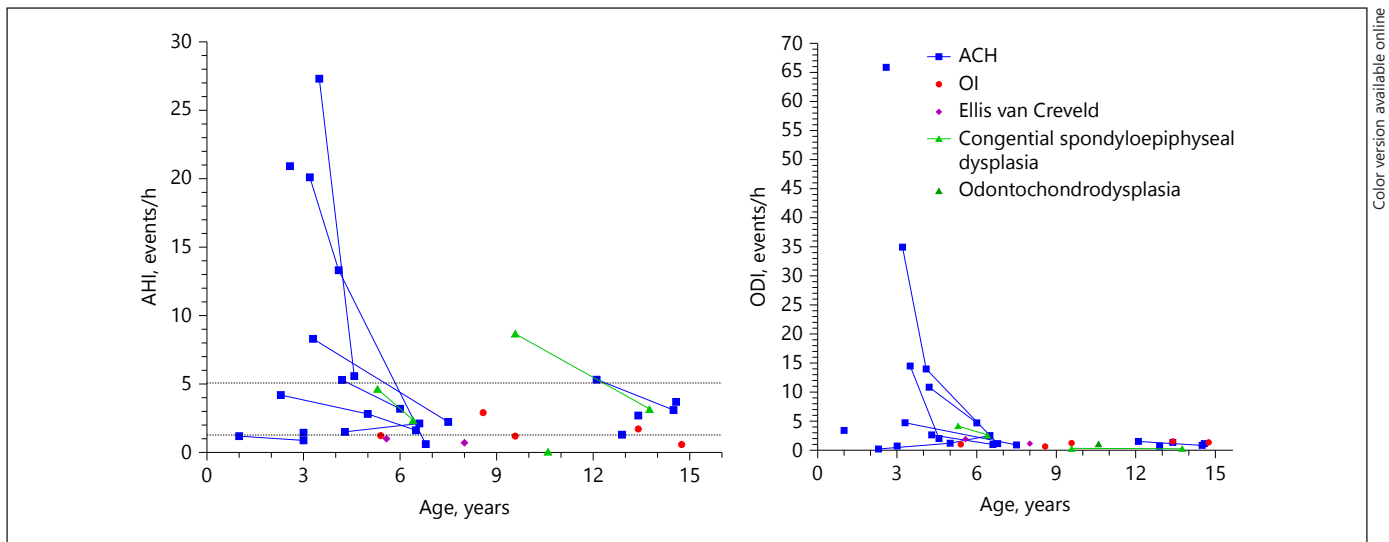


Fig. 1. Plotted distribution of the AHI (events/h) and ODI (events/h) values of the population study (ACH and variants, OI and Ellis van Creveld syndrome), according to age at measurement. ACH, achondroplasia; OI, osteogenesis imperfecta; AHI, apnea-hypopnea index; ODI, oxygen desaturation index.

Table 1 shows the results of polygraph studies on ACH children. ACH children who had been both surgically and medically treated had a lower mean AHI (2.1 ± 1.1 vs. 3.2 ± 1.9 events/h) than untreated ACH children (baseline, 9.5 ± 9.6 events/h). At follow-up, the ACH children who had been medically treated had higher mean values

than those who had been surgically treated. Because of the low number of patients, none of the comparisons were statistically significant ($p > 0.05$). After medical and surgical interventions, the number of children with ACH with normalization of AHI was 1 out of 6 and 1 out of 7, respectively.

Table 2. Results of polygraph study in patients with congenital spondyloepiphyseal dysplasia ($n = 2$), odontochondrodysplasia ($n = 1$), osteogenesis imperfecta ($n = 6$), and Ellis van Creveld syndrome ($n = 2$)

Syndromes	Spondyloepiphyseal dysplasia				Odonto-chondro-dysplasia	Osteogenesis imperfecta type III		Osteogenesis imperfecta type IV	Osteogenesis imperfecta type VI			Ellis van Creveld	
	1		2		1	1	2	3	4	5	6	1	2
Timing	T0	T1	T0	T1	T0	T0	T0	T0	T0	T0	T0	T0	T0
Age, years	5.3	6.4	9.6	13.8	10.6	13.4	12.1	5.4	14.8	9.6	8.6	5.6	8
Age at surgery, years	2	–	4.5	–	–	–	–	–	–	–	–	–	–
eTST, h	10	10.2	9.1	7.9	9.3	8	7.9	8	8.3	9.3	9.5	10.7	9.5
OA, events/h	1.2	0	5.3	0.6	0	0	0.1	0	0	0.2	1.4	0	0
CA, events/h	0.6	0.3	2.9†	2.1†	0	1.5	0	0.4	0.3	0.4	1.1	0	0.4
H, events/h	2.9	2.1	0.4	0.1	0.1	0.1	0.1	0.4	0.4	0.5	0.1	1	0.3
AHI, events/h	4.7	2.4	8.7	3.2	0.1	1.7	0.2	1.2	0.6	1.2	2.9	1	0.7
ODI, events/h	4.2†	2.6†	0.4	0.3	1.1	1.5†	0	1	1.3†	1.2	0.6	1.9†	1.1
Mean SpO ₂ , %	96	99	98	97	96	98	98	97	98	97	98	95	98
Min SpO ₂ , %	88	74	80	94	90	92	96	90	93	91	93	90	90
Snoring (% eTST)	0.1	0	34	36.3	0	0	0	0.6	0	0.2	0	7	0.1

Children with achondrodysplasia and congenital spondyloepiphyseal dysplasia received a follow-up evaluation (T1) after baseline evaluation (T0). “†” indicates increased values according to normal values reported for age [23].

AHI, apnea-hypopnea index; H, hypopnea; OA, obstructive apnea; CA, central apnea; ODI, oxygen desaturation index; OI, osteogenesis imperfecta; eTST, estimated total sleep time.

Table 2 shows the baseline (T0) and follow-up (T1) sleep respiratory parameters of the two children with spondyloepiphyseal dysplasia, who had adenotonsillectomy before the admission in the study. In both children, mild and moderate sleep respiratory involvement was found. They were medically treated and both showed amelioration of the AHI at follow-up. Table 2 shows the results of overnight analysis of subjects with OI and EVC. Mild respiratory involvement was present in 4/6 patients with OI. One of two patients with EVC syndrome had borderline mild sleep respiratory involvement (AHI: 1 event/h) and increased ODI (1.9 events/h) in comparison with normal values for age [23].

Magnetic Resonance Imaging

Table 3a shows the results of the MRI measures (mm³ and %) of the nasopharynx, oropharynx, and hypopharynx spaces and the sleep respiratory polygraph results (AHI, obstructive apneas, hypopneas [H], and ODI) in 11 ACH children. In one child (No. 6), adenotonsillectomy was done after MRI and before the polygraph study. MRI was not available in two children with ACH.

Table 3b shows the correlation between polygraph results (AHI and ODI) and MRI measures (% of totals). Despite the low number of measurements, a negative

correlation was found between sleep respiratory patterns (AHI, OI, H) and nasopharynx plus oropharynx space (%). This might suggest that the smaller the nasopharynx space, the greater the sleep respiratory disturbance.

Discussion

None of our subjects with ACH had cervicomedullary complications. We observed the presence of moderate-severe OSAS in 60% of ACH children at enrollment. At follow-up, patients who had A&T also showed lower OSAS severity due to the fact that adenotonsillectomy was effective in gaining some improvement in sleep respiratory disturbances; adenotonsillectomy should be considered for treatment in children with severe ACH. The reduced prevalence of OSAS after adenotonsillectomy supports the benefit of surgery [26]. Sleep respiratory disturbances decreased with age in those who did not perform adenotonsillectomy, and this could be related to physiological involution of the nasopharynx lymphoid tissue [27]. Interestingly, correlation between MRI and respiratory parameters showed that sleep respiratory disturbances correlated negatively with relative nasophar-

Table 3.

a. Results of the MRI measures concerning nasopharynx, oropharynx, and hypopharynx spaces (mm³ and % total volumes; see also Figure 1) and sleep polygraph measures (AHI, OA, H, and ODI) in children affected by ACH. Patient No. 6 underwent adenotonsillectomy after MRI and before the sleep polygraph study

Patient ID number	Magnetic resonance imaging						In-laboratory overnight respiratory polygraphy					
	age, years	nasopharynx space		oropharynx space		hypopharynx space		age, years	AHI, events/h	OA, events/h	H, events/h	ODI, events/h
		mm ³	%	mm ³	%	mm ³	%					
6	0.71	202	7.39	1,083	39.61	1,449	53.00	4.3	1.5	0.5	0.9	2.6
1	2.08	1,915	36.16	1,792	33.84	1,589	30.00	1.0	1.2	0.0	1.2	3.4
9	2.39	809	20.97	1,083	28.07	1,966	50.96	2.0	2.3	2.1	0.2	0.3
5	2.98	1,372	26.22	2,488	47.55	1,372	26.22	5.3	4.7	1.2	2.9	4.2
7	4.68	203	5.31	969	25.33	2,653	69.36	4.6	72.0	31.5	34.2	57.1
3	4.79	2,099	34.10	1,713	27.83	2,343	38.07	4.2	5.3	0.0	4.2	10.8
2	5.87	1,054	17.26	3,002	49.16	2,051	33.58	4.1	13.3	5.4	5.5	14.0
11	8.85	977	36.90	765	28.89	906	34.21	12.1	0.2	0.0	0.2	0.2
8	12.19	2,850	51.71	1,416	25.69	1,246	22.61	12.1	5.3	2.5	1.0	1.5
4	12.93	1,723	18.18	2,525	26.64	5,229	55.18	12.9	1.3	0.0	0.6	0.7
10	13.42	2,775	36.12	2,490	32.41	2,418	31.47	13.4	2.7	1.6	0.2	1.3

b Correlation analysis (Pearson's *s*) between sleep respiratory results (AHI, OA, H, and ODI – events/h) and relative (%) nasopharynx, oropharynx, and hypopharynx spaces measured by brain MRI analysis of 10 achondroplasia patients. For the execution of the statistical analysis, we excluded patient No. 6 (see panel A)

Volumes, %	AHI <i>p</i> value (<i>r</i>)	OA <i>p</i> value (<i>r</i>)	H <i>p</i> value (<i>r</i>)	ODI <i>p</i> value (<i>r</i>)
Nasopharynx	0.115 (–0.502)	0.113 (–0.505)	0.106 (–0.514)	0.102 (–0.519)
Oropharynx	0.548 (–0.204)	0.521 (–0.217)	0.567 (–0.194)	0.652 (–0.154)
Hypopharynx plus oropharynx	0.048 (–0.606)	0.044 (–0.616)	0.044 (–0.613)	0.054 (–0.594)

AHI, apnea-hypopnea index; H, hypopnea; OA, obstructive apnea; ODI, oxygen desaturation index.

ynx plus oropharynx space. We speculate that the smaller relative nasopharynx plus oropharynx space had the higher number of obstructive events during sleep. MRI could predict the risk of obstruction of upper airways in patients with ACH because certain skeletal pattern is important in the presence and progression of the obstruction. In conclusion, MRI could be an accurate technique evaluating the prevalent risk in children with the risk of developing OSAS [28].

Further, we performed a sleep respiratory study in two patients with spondyloepiphyseal dysplasia, with moderate OSAS at baseline, and mild OSAS during follow-up; both had adenotonsillectomy before the polygraph test. With a respiratory pattern similar to subjects with ACH, the patient with odontochondrodysplasia showed normal sleep respiratory pattern.

A lower prevalence of OSA after adenotonsillectomy has been reported, although in several cases the clinical

problems remained [26]. Obstruction of upper airways could not always be related only to adenotonsillar hypertrophy, but also to mid-face hypoplasia and upper airway soft tissue abnormalities, and various treatments have been suggested [6]. Thus, persisting mild OSAS in several of our subjects with ACH, even after adenotonsillectomy, indicates that further treatment may be warranted. Orthodontic treatment (i.e., rapid maxillary expansion) could be another approach that increases treatment efficacy [29].

The majority of our subjects with OI showed mild sleep respiratory disturbances, whereas significant desaturations according to the ODI references were reported [23]. One of our patients with OI type VI showed mild OSAS in which the obstructive item was prevalent, but to our knowledge, no literature data on sleep respiratory investigation in OI children are available. Our investigation was done because they have a unique facial profile and malocclusions become stronger with increasing age. Our

OI patients have retarded vertical dimensions, flattened cranial base angle, relative prognathic, larger facial divergence, and more forward counterclockwise mandibular growth [30]. The OI type III patients were characterized by a paradoxical inspiratory inward motion of the pulmonary rib cage, significant thoraco-abdominal asynchronies, and rib cage distortions in a supine position [31]. Our results confirm that OI had increased mean thoracoabdominal asynchrony in the supine position in comparison with children with ACH. These findings suggest a paradoxical thoracoabdominal movement and possible continuous partial obstruction of the upper airway. Studies have shown the presence of OSA in children OI and adults secondary to laryngomalacia or redundant supra-glottic or epiglottis mucosa [32].

In our study, we found a mild respiratory disturbance in one out of two EVC patients, related to H, and an increased ODI. We were unable to find any published data on sleep respiratory disturbance in such patients. Hypoplasia of the anterior maxilla, prognathic of the mandible and the increased height of the lower third of the face were previously described [33]. In children with EVC, the prognosis is mainly associated with respiratory difficulties in the first months of life [17].

The main limitation of our study is that it is retrospective, although the major strength of this study is the assessment of rare under-reported skeletal syndromes, and our findings could influence more research on these syndromes. Another strength of our study is that the patients were examined for daily clinical practice, limiting

the time of their stay in the hospital. In fact, they were admitted in the evening, stayed overnight in a quiet, properly prepared sleep chamber, and then dismissed the next morning at the end of their clinical assessment and treatment.

Conclusions

The in-laboratory respiratory polygraph study was helpful in screening for OSAS in our subjects with rare skeletal diseases. Relevant sleep respiratory disturbances were detected in ACH, as well as in OI and EVC Syndrome. However, a negative correlation was observed between sleep respiratory patterns and nasopharynx plus oropharynx space assessed by MRI. We speculate that smaller the nasopharynx space, the greater the sleep respiratory disturbance. Adenotonsillectomy was successful in patients with ACH. Multicenter studies are needed to add information on sleep respiratory patterns in these under-reported skeletal diseases.

Disclosure Statement

None of the authors declared a conflict of interest.

Funding Sources

No financial support was received for this study.

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