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Obstructive Sleep Apnea in Adults with Treacher Collins Syndrome is Related with Altered Anthropometric Measurements, Increased Blood Pressure and Impaired Quality of Life

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Abstract

Objectives This study aimed at evaluating the risk for obstructive sleep apnea (OSA) and its frequency in adults with Treacher Collins syndrome (TCS). The association of OSA with excessive daytime sleepiness (EDS), respiratory symptoms, and clinical variables was also assessed.

Material and Methods The subjects were prospectively screened for OSA through the Berlin Questionnaire and type I polysomnography. The Epworth Sleepiness Scale and the Respiratory Symptoms Questionnaire were used for assessing OSA-related symptoms. Quality of life was assessed by means of the Short Form 36 Health Survey.

Results The sample comprised 20 adults with TCS (55.0% female), aged 22.6 ± 5.8 years. Mean values of systemic blood pressure ($113.0 \pm 12.6/68.0 \pm 9.5$mmHg), body mass index ($22.9 \pm 5.9$kg/m^2), neck ($34.1 \pm 4.3$cm), and waist circumference (80.4 ± 13.6cm) characterized the sample. A high risk for OSA was detected in 35% of the sample. Polysomnography results indicated an OSA frequency of 44.4%, with a median apnea-hypopnea index (AHI) value of 3.8 events/hour (minimum = 0.2; maximum = 77.5). Snoring (75.0%), nasal obstruction (70.0%) and EDS (20.0%) were the reported OSA-related symptoms. Quality of life median scores were 72.3 points (minimum = 45.0; maximum = 91.1). Strong positive correlations between AHI versus waist circumference and AHI versus systolic blood pressure were found. Moderate positive correlations between AHI versus body mass index and AHI versus neck

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- mandibulofacial dysostosis
- airway obstruction
- polysomnography

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circumference were detected. Negative correlation between AHI versus vitality were also observed.

Conclusion Adults with TCS are at high risk for OSA, which is associated with respiratory symptoms, altered anthropometric measurements, increased systolic pressure and impairment of quality of life.

Introduction

Treacher Collins syndrome (TCS) is a rare craniofacial anomaly that belongs to the group of mandibulofacial dysostoses.[1] It occurs with an estimated incidence of ~1 in 80,000 births and 60% of the cases result from de novo mutations.[2,3] This syndrome develops solely from abnormal differentiation of the first and second pharyngeal arches,[3] causing a cluster of bilateral temporoaural malformations, zygomatic and mandibular dysplasia.[4]

Variability of head and neck structure's malformations in TCS results in a heterogeneous phenotype. Bilateral microtia, atresia, or stenosis of the external auditory canal, middle ear cavity hypoplasia, and dysmorphic ossicular chain cause bilateral hearing loss.[5] Skeletal dysmorphism of the orbits, with malar hypoplasia, results in characteristic alterations, such as downslanting palpebra and fissure narrowing sign, eyelid coloboma, and absence of the inferior lacrimal puncta.[6] Major salivary gland hypoplasia or aplasia, ensuing hyposalivation, negatively affects speech, mastication, and deglutition – and increases the risk for stomatological and dental pathologies.[7]

Narrowing of the upper airway accounts for impaired normal ventilation during sleep,[8] leading to a high prevalence of breathing disorders in individuals with TCS, including obstructive sleep apnea (OSA).[9,10] This sleep-disordered breathing that is common among patients with craniofacial anomalies,[11] arises as result of several factors, including, presence of cleft palate, and palatoplasty,[12] choanal atresia or stenosis,[3] and pharyngeal collapse due to mandible hypoplasia and retrognathia.[13] The recurrent episodes of upper airway collapse seen in OSA, cause apneic events with hypoxia, symphatetic activation followed by increased pulse and blood pressure, arousals for re-oxigenation and release of inflammatory factors.[14]

Strong evidences recommend that OSA prediction in adults should include in-laboratory polysomnography or at-home sleep apnea testing.[8] However, in non-sleep clinic settings, questionnaires and morphometric models may be helpful to identify individuals who are at risk for this sleep-disordered breathing.[8] Studies that conjointly applied these methods for OSA diagnosis among adults with TCS, are scarce.[10,15,16] However, due to deleterious effects of undiagnosed OSA, evidences in this field are of great relevance.[8,10,16–18]

Early diagnosis of OSA is mandatory, since it is a symptomatic disorder that predisposes for cardiometabolic complications,[17] affects quality of life,[10] causes drowsiness, and decline of motor and cognitive performance.[18] Therefore, this study aimed at evaluating OSA frequency in adults with TCS, verifying its severity and relationship with anthropometric measurements, excessive daytime sleepiness, respiratory symptoms and quality of life.

Material and Methods

Study Design and Bioethical Considerations

This is a descriptive study that prospectively evaluated adults with a confirmed diagnosis of TCS, which were undergoing routine clinical evaluation. Calculation of the sample size was done considering a population of 76 individuals with TCS ($\geq$16 years) currently registered at a reference center for patients with craniofacial anomalies. An expected mean OSA frequency of 87% was adopted, as described for adults with TCS in the study of Østertun Geirdal et al.[10] A sample of 16 individuals was calculated to predict an 87% $\pm$ 15 OSA frequency, with a 95% confidence level (α 0.05) (OpenEpi v. 3.01).

During an eight-month period, 22 patients with TCS aged $\geq$ 16 years were scheduled for ambulatory follow-up and therefore invited to participate. The acceptance rate was 90.9%, resulting in a sample of 20 adults. None carried severe or uncontrolled physical or mental health conditions that would interfere with this study's purpose.[10] The study protocol was approved by the Institutional Review Board (process number 85275618.0.0000.5441) and complied with the Declaration of Helsinki. All the participants gave their informed consent prior to their inclusion in the study.

Clinical Evaluation

Demographic information and history of interventions involving the upper airway, were obtained from patient's charts, aiming the sample characterization.[10,16] Physical examination included assessment of body mass index (BMI, kg/m^2),[19] measurements of neck and waist circumferences (cm),[20,21] and systemic blood pressure (mmHg) evaluation.[22]

Questionnaires

The Berlin Questionnaire[23] assessed the risk for having OSA. This instrument consists of eleven items, gathered into three different domains, aiming at classifying the patient as at high or low risk for OSA. Sensitivity and specificity of this instrument for the identification of an $\geq$ 5 apnea hypopnea index (AHI) cutoff is 0.76 and 0.45, respectively.[8]

The Epworth sleepiness scale (ESS),[24] provided a measurement of the subject's general level of daytime sleepiness. This scale is based on eight soporific situations, giving a score between 0 and 24 (excessive daytime sleepiness $\geq$ 10). Sensitivity and specificity of the ESS for the identification of an AHI $\geq$ 5 is 0.76 and 0.45, respectively.[8]

A Respiratory Symptoms Questionnaire adapted from Caouette-Laberge et al.,[25] was used to evaluate respiratory distress over the day- and night-times. Self-perceived symptoms were assessed as dichotomic variables (No / Yes).

A generic measure of health status, the 36-item Short-Form Health Survey (SF-36)[26] provided subjective measures of physical function and dysfunction, and mental distress and well-being. In a scale between 0 and 10, a cutoff value < 40 defines poor quality of life.

In-laboratory Sleep Study

Complete in-laboratory nocturnal type I polysomnography was performed using the EMBLA-N7000 digital system (Embla Systems, Inc., Broomfield, CO). Recordings of electroencephalogram (F3-A2, F4-A1, C3-A2, C4-A1, O1-A2, O2-A1), electrooculogram (two channels), chin and anterior tibialis electromyogram, electrocardiogram, oronasal airflow (by thermistor and nasal pressure cannula), abdominal and thoracic movements, body position, oxygen saturation, and heart rate, were provided.[27]

OSA was classified according to the American Academy of Sleep Medicine (AASM) criteria.[8] Apnea-hypopnea index values ≥ 5 events per hour of sleep were considered indicative of OSA. OSA severity was classified as mild (AHI = 5–14.9/h), moderate (AHI = 15–29.9/h), or severe (AHI $\geq$ 30/h).

Data Analysis

Shapiro-Wilk test was used for assessing normality. Continuous data were presented as mean $\pm$ standard deviation, or as median values and their 25% and 75% percentiles, when appropriate. Frequency and percentages described categorical variables. Fisher's exact test was applied for the analysis of contingency tables. The bivariate statistical model, Spearman's rank correlation coefficient was used to test associative hypothesis. The level of statistical significance was set as $p \leq 0.05$.

Results

Clinical Evaluation

The sample comprised 20 subjects, 55.0% female, with a mean age of 22.6 $\pm$ 5.8 years (16–35 years of age), which not differed between sexes ($p > 0.05$). Distinctive abnormalities of TCS, especially those involving the zygomatic complex and the mandible, were highly prevalent (70.0%) (►**Fig. 1A**, **1B**).

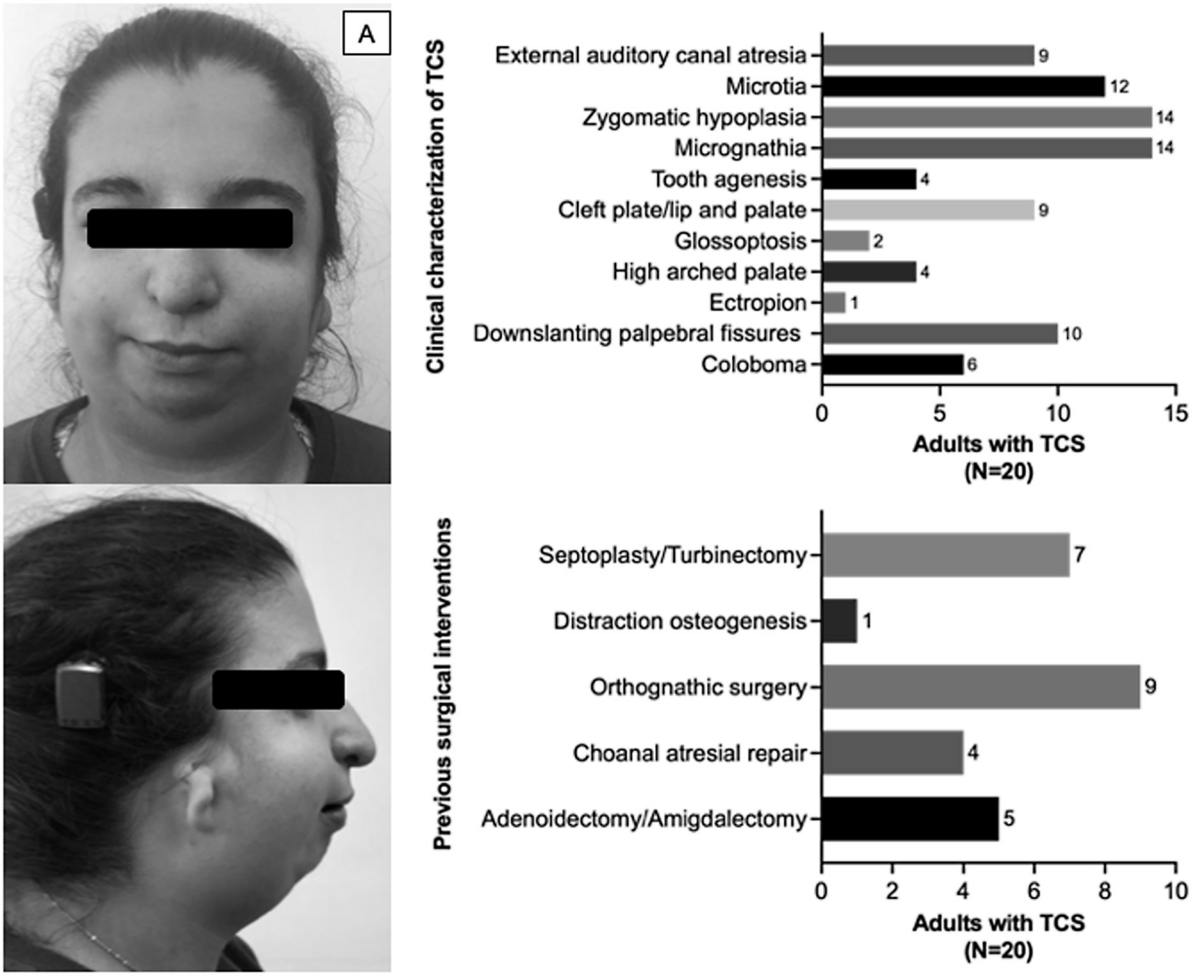

Fig. 1 Summarization of clinical assessment and charts review findings: (**A**) frontal and lateral aspect of a female subject with distinctive abnormalities of Treacher Collins syndrome (TCS) - microtia, downslanting palpebral fissure, zygomatic and mandibular hypoplasia.

Table 1 Anthropometric and systemic blood pressure measurements in adults with Treacher Collins syndrome (N = 20).

Variables	Total (N = 20)	Male (n = 9)	Female (n = 11)
	Mean ± SD	Mean ± SD	Mean ± SD
Body mass index (kg/m^2)	22.90 ± 5.90	22.63 ± 4.27	23.13 ± 7.18
Neck circumference (cm)	34.05 ± 4.29	36.89 ± 2.93^A	31.73 ± 3.87^A
Waist circumference (cm)	80.40 ± 13.63	84.44 ± 10.73	77.09 ± 15.31
Systolic blood pressure	113.00 ± 12.61	121.10 ± 11.67^B	106.40 ± 9.24^B
Diastolic blood pressure	68.00 ± 9.51	71.11 ± 10.54	65.45 ± 8.20

Notes: Data passed Shapiro-Wilk normality test. Same letters indicate differences between mean values, at a statistical level of significance (Student's t test, $p \leq 0.05$).
Abbreviations: N, sample size; n, group size; SD, standard deviation.

Previous medical history of interventions involving the upper airway (►**Fig. 1C**), showed that orthognathic surgery was the most frequently performed (45.0%).

Clinical data (anthropometry and systemic arterial pressure) are presented in ►**Table 1**. Considering provided reference values,[19–21] the evaluated adults with TCS were eutrophic, with BMI and neck and waist circumference within expected parameters. Males had higher values of systolic blood pressure (120–129 mm Hg) than females, and 3 cases of stage 1 hypertension (130–139 mm Hg)[22] were diagnosed among them.

Questionnaires

Results of the Berlin Questionnaire assessment indicated high risk for OSA in 35% (7) of the sample. There was no association between male sex, or overweight, and high risk for OSA ($p > 0.05$).

Excessive daytime sleepiness (global score > 10) was identified in 20.0% of the participants. Snoring (75.0%) and nasal obstruction (70.0%) were the most prevalent findings in the Respiratory Symptoms Questionnaire (►**Fig. 2**–A). Quality of life evaluated through the SF-36 instrument revealed a median global score of 72.3 points (25% percentile = 65.2; 75% percentile = 82.0). Lower median scores (< 70) were found in the domains of role emotional, vitality, general and mental health, without differences between male and female ($p > 0.05$) (►**Fig. 2**–B). Poor quality of life defining cutoff values (< 40) were not reached for any subject, considering the resultant mean scores of the assessed domains.

The distribution of individuals with high risk for OSA and excessive daytime sleepiness, snoring, nasal obstruction and without previous history of orthognathic surgery is shown in ►**Fig. 3**–A to D. It was observed that 100.0% (7) of snorers and 87.1% (6) of those who were not submitted to orthognathic surgery, had a subjective higher risk for OSA. Nonetheless statistically significant associations were not found ($p > 0.05$).

In-laboratory Sleep Study

Of the whole sample, 9 (45%) subjects accepted to undergo an in-laboratory sleep test. A definitive diagnosis of OSA was given to 4 (44.4%) individuals, of whom 3 (33.3%) had mild/moderate OSA, and 1 (11.1%) presented the severe form of the disease. Median values of AHI were 3.8 events/hour, with minimum and maximum values of 0.2 and 77.5, respectively. The minimum oxygen saturation median value was 94.0% (minimum = 58.0%; maximum = 97.0%). ►**Table 2** presents descriptive statistics of the assessed parameters through type 1 polysomnography.

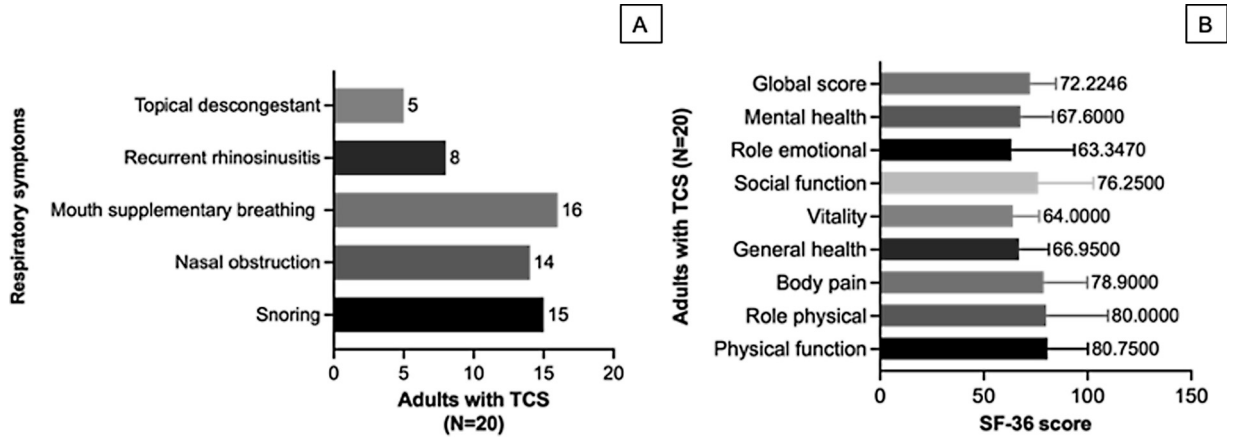

Fig. 2 Descriptive results of the Respiratory Symptoms Questionnaire (**A**) and median scores of the 36-item Short-Form Health Survey (SF-36) (**B**).

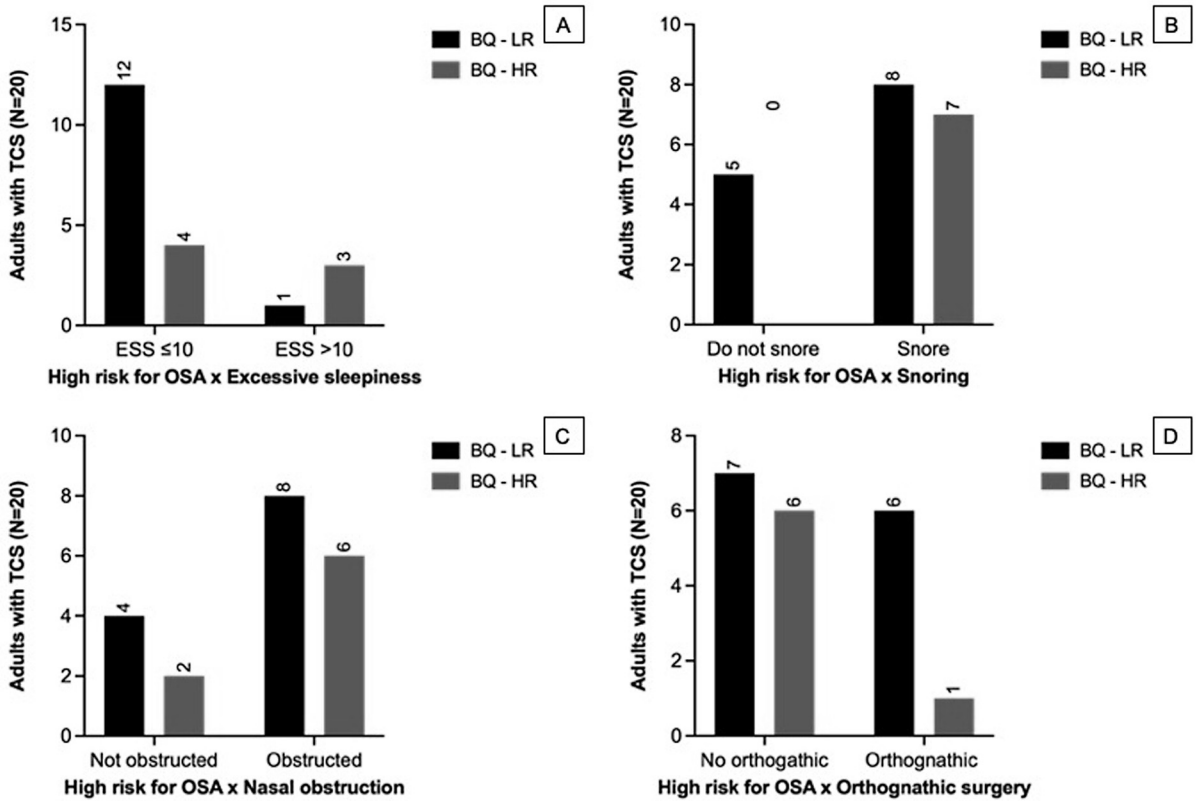

Fig. 3 Sample distribution considering the subjective assessment of risk for obstructive sleep apnea through the Berlin Questionnaire (BQ) considering excessive daytime sleepiness (**A**), snoring (**B**), nasal obstruction (**C**), and history of a previous orthognathic surgery (**D**).

No statistically significant associations between OSA severity with categorical data obtained through questionnaires were detected. The distribution of participants concerning the subjective risk for OSA, excessive daytime sleepiness, snoring, nasal obstruction, and orthognathic surgery among the objective ranges of AHI (events/hours), is summarized in ►**Table 3**. The only subject with severe OSA (AHI = 77.5 events/hours; SaO_2 = 58.0%), presented a high risk for OSA in the Berlin Questionnaire, snoring and had no history of orthognathic surgery, although had normal daytime sleepiness and no nasal obstruction.

Table 2 Characterization of polysomnographic findings in adults with Treacher Collins syndrome (N = 9)

Parameters	Median	25% percentile	75% percentile	Min	Max
Arousals (n°)	21.00	15.50	62.00	0.00	187.00
Arousals index (events/hour)	5.30	3.50	9.65	0.00	31.10
Apneas + Hypopneas (n°)	22.00	10.00	43.00	1.00	629.00
Hypopneas (n°)	8.00	2.50	17.00	0.00	36.00
Obstructive apneas (n°)	10.00	2.50	21.50	1.00	627.00
Central apneas (n°)	0.00	0.00	1.00	0.00	4.00
Mixed apneas (n°)	0.00	0.00	0.00	0.00	5.00
Apnea-hypopnea index (events/hour)	3.80	1.45	6.65	0.20	77.50
REM apnea-hypopnea index (events/hour)	4.50	1.00	12.00	0.60	87.70
Duration of apnea and hypopnea (seconds)	40.50	27.30	52.05	17.20	78.40
Mean SaO_2 (%)	97.00	96.40	98.05	86.90	99.00
Minimum SaO_2 (%)	94.00	88.50	95.00	58.00	97.00

Notes: Data not passed Shapiro-Wilk normality test.
Abbreviations: n°, number; Min, minimum; Max, maximum; REM, rapid-eye-movement sleep; SaO_2, oxygen saturation.

Table 3 Descriptive tabular representation of categorical data obtained through questionnaires, according to the severity of objectively diagnosed obstructed sleep apnea in adults with Treacher Collins syndrome (N = 9)

		Obstructive sleep apnea (AHI events/hour)		
		None < 5 (n = 5)	Mild/Moderate ≥ 5 to < 30 (n = 3)	Severe ≥ 30 (n = 1)
Berlin Questionnaire (risk for OSA)	Low	4	1	0
	High	1	2	1
Epworth Sleepiness Scale (daytime sleepiness)	Normal	4	2	1
	Excessive	1	1	0
Snoring	No	1	1	0
	Yes	4	2	1
Nasal obstruction	No	1	1	1
	Yes	4	2	0
Orthognathic surgery	No	3	2	1
	Yes	2	1	0

Notes: AHI <5 = no obstructive sleep apnea; ≥ 5 AHI ≤ 30, mild to moderate obstructive sleep apnea; AHI >30, severe obstructive sleep apnea.
Abbreviations: OSA, obstructive sleep apnea; AHI, apnea-hypopnea index (events/hour).

Positive correlations between AHI versus neck (r = 0.5) and waist (r = 0.8) circumferences, body mass index (r = 0.5), and systolic blood pressure (r = 0.8) were detected. Oppositely, negative correlations were found between AHI versus social function (r = 0.8), physical activity (r = 0.5), and vitality (r = 0.6) (►**Table 4**). Continuous scores of the Epworth Sleepiness Scale did not correlated with polysomnography results.

Discussion

Descriptive studies aiming at characterizing OSA prevalence and associated risk factors among adults with TCS are still scarce (►**Table 5**), despite their relevance for evidence-based clinical management of these patients. The present study evaluated adults aged 16 to 35 years with TCS, combining survey methods, anthropometric assessment and laboratory analysis. The main findings were a high risk for OSA identified in 35% of the sample and a definitive diagnosis of this sleep-disordered breathing through gold-standard criteria resulting in a 44.4% frequency.

When cases were individually evaluated, the Berlin Questionnaire presented a false negative rate of 25.0% for the AHI ≥ 5 cutoff. Therefore, in this particular group of 4 subjects with TCS and OSA, this instrument displayed a limited diagnosis utility.[8] Indeed, according to the literature, there is a discrepancy between the manifested symptoms and OSA occurrence, which can result in underdiagnosed cases of this condition.[16] In this context, the Berlin Questionnaire can be considered an important tool for OSA screening in the clinical setting, contributing to the early identification of those most in need to be referred for in-laboratory sleep tests. Presently, a screening tool specific to the craniofacial population is being developed by our group.

The objective prevalence rates of OSA in this study were similar to that found by Plomp et al.[16]–44.4% and 41.0%, respectively. Mild/moderate cases were predominant in the present sample, while severe cases were most prevalent in the sample of Plomp et al.[16] Such differences are attributed to the wide age range of individuals with TCS assessed by them, including middle-aged and elderly subjects, which are more prone to have the severe form of OSA.[28]

Akre et al.[15] and Østertun Geirdal et al.[10] detected OSA rates among adults with TCS that practically double ours – 100.0% and 87.0%, respectively. In the present study, there was no exclusion of patients submitted to surgical procedures that would be affected the upper airway's patency, as seen in Plomp et al.[16] One could consider this a limitation of the present study, contributing to a lower OSA prevalence as detected by Akre et al.[15] and Østertun Geirdal et al.[10]

On the other hand, the authors understand that including patients previously submitted to airway surgeries constitute a marginal limitation of this study. Mainly because adults with TCS have upper airways that are multisite obstructed, and associated with several craniofacial malformations.[3,12,13,16] Therefore, isolated surgical procedures are unlikely to promote long-standing clinical resolution or the mitigation of OSA severity.[9,16] Enlightening the influence of airway procedures over OSA occurrence and severity should require pre- and post-operative studies and longitudinal assessments.

Remarkably, the studied sample presented a 15% higher frequency of OSA than young adults (aged 20–29) with cleft lip and palate (29%) – the most prevalent craniofacial anomaly.[29] Also, OSA prevalence among those with TCS was nearly 30% and 40% greater than males (12.4%) and females (1.7%) (aged 20–29) without craniofacial anomalies, respectively.[28] In the clinical context, these findings highlight the

Table 4 Spearman's Rho values showing correlations between polysomnography parameters vs. anthropometric measurements, blood pressure data, and quality of life (N = 9)

	NC	WC	SBP	DBP	Ar	A + H	H	AO	AHI	AHI-REM	SaO_2 min	dAH	PA	Pain	Vitality	SF
BMI	0.61	0.68	0.22	0.34	−**0.70**	0.59	0.20	0.58	0.53	**0.89**	−0.37	0.60	0.08	−0.19	−0.25	−0.25
NC		**0.80**	0.69	0.34	−0.44	0.66	0.06	0.65	0.50	**0.75**	−0.65	0.51	−0.21	−0.10	−0.43	−0.04
WC			0.61	0.46	−0.27	**0.85**	0.24	0.71	**0.79**	0.69	−0.66	0.62	−0.26	−0.23	−0.39	−0.29
SBP				0.47	−0.19	**0.89**	0.42	0.73	**0.79**	0.51	−0.58	**0.70**	−0.36	0.04	−0.48	0.09
DBP					0.17	0.28	0.64	−0.04	0.43	0.04	0.27	**0.71**	−0.01	−0.15	−0.60	−0.07
Ar						−0.15	**0.81**	−0.28	−0.04	−0.56	0.24	−0.35	−0.36	0.40	0.06	0.35
A + H							0.40	**0.91**	**0.95**	0.58	−**0.72**	0.59	−0.57	−0.62	−0.59	−0.68
H								0.17	0.44	−0.38	0.08	0.43	−**0.70**	−0.14	−0.57	−0.19
AO									**0.78**	0.54	−**0.88**	0.30	−0.44	−0.42	−0.24	−0.46
AHI										0.46	−0.53	0.62	−0.50	−**0.71**	−0.64	−**0.78**
AHI-REM											−**0.72**	0.54	0.33	0.10	−0.36	−0.30
SaO_2 min												−0.10	0.21	0.24	0.06	0.19
dAH													−0.32	−**0.74**	−0.92	−0.68
PA														0.19	0.34	0.38
Pain															0.50	0.50
Vitality																0.50

Notes: Values indicate strong ($r \geq 0.70$) and moderate ($r \geq 0.40$) correlations between variables at a statistically significant level ($p \leq 0.05$).
Abbreviations: BMI, body mass index (kg/m^2); NC, neck circumferences (cm); WC, waist circumferences (cm); SBP, systolic blood pressure (mmHg); DBP, diastolic blood pressure (mmHg); Ar, arousals (n°); A + H, Apneas + Hypopneas (n°); H, hypopneas; OA, obstructive apneas; AHI, apnea-hypopnea index(events/hour); AHI-REM, rapid-eye-movement sleep apnea-hypopnea index(events/hour); SaO_2 min, minimum oxygen saturation (%); dAH duration of apnea and hypopnea (seconds); PA, physical activity.

Table 5 Definitive diagnosis of obstructive sleep apnea frequency in adults with Treacher Collins syndrome, according to this and previous studies

Studies	Sample size	Age (years)	OSA occurrence (%)	AHI $\geq$ 5 (%)	AHI $\geq$ 30 (%)
Present study	9	22.7 $\pm$ 6.6	44.4	75.0	25.0
Østertun Geirdal et al.[10]	15	38.6 $\pm$ 18.5	87.0	46.7	53.3*
Akre et al.[13]	11	47.2 $\pm$ 13.2	100.0	63.7	36.4
Plomp et al.[14]	22	37 (20 - 60)	41.0	33.3	66.7

Notes: Sample sizes of groups having type 1 polysomnography, and not including infants or children, according to different criteria defined by the authors; AHI $\geq$ 5 = mild/moderate OSA; AHI $\geq$ 30 = severe OSA; *AHI > 15 = moderate/severe OSA.
Abbreviations: PSG, polysomnography; AHI, apnea-hypopnea index(events/hour).

importance of preforming OSA screening in all individuals diagnosed with TCS.

Anthropometric measures are risk factors linked with the onset of sleep-disordered breathing,[30] cardiovascular diseases,[31] and metabolic syndrome.[32] Apnea-hypopnea index and OSA-defining anthropometric measures were variables that moved in tandem, showing a positive correlation in adults with TCS. Previous studies did not report this relationship in other samples of adults with TCS.[10,15,16] The association between anthropometric measurements and OSA occurrence – although not posing a cause-effect relation – indicates that additional factors than craniofacial characteristics should be carefully followed-up in this population.[3,12,13]

Another original finding of this study compared with others[10,15,16] was that blood pressure positively correlated with several polysomnography variables (number and duration of apneas and hypopneas, obstructive apneas, AHI and AHI during rapid-eye-movement sleep, and minimum oxygen saturation). In the evaluated sample, male subjects accounted for 3 cases of stage 1 hypertension (130–139 mm Hg). Male sex and OSA are relative risk factor for essential and medication-resistant hypertension.[33] High blood pressure – especially systolic as seen in this study – is associated with a higher risk for coronary disease, stroke and chronic renal disease.[34] Therefore, this is a finding that recommends further efforts for awareness and prevention hypertension, and prospective systemic blood pressure evaluation among young adults with TCS, as part of their long-term clinical follow-up.

Excessive daytime sleepiness evaluated through the Epworth Sleepiness Scale was not a prominent finding among our sample (20%). This finding was unexpected, since drowsiness is a well-known consequence of OSA. Nevertheless, our sample presented a 7% higher frequency of excessive sleepiness than Østertun Geirdal et al.[10] reported earlier (13%). The literature indicates that scores of this scale, did not predict well polysomnographic values.[35] Likewise, limited sensitivity (0.3–0.7), specificity (0.5–0.8), and accuracy (51.0–59.0%) for the AHI $\geq$ 5 cutoff were described.[8] Indeed, no correlation was found between this instrument and polysomnographic variables in the present study.

Snoring is one of the cardinal symptoms of OSA and a high frequency of this condition was observed in the present sample (75.0%), certainly representing a burden among the evaluated subjects. However, no associations of snoring with an AHI $\geq$ 5 or AHI $\geq$ 30 were found, as reported by Akre et al.[13] Patients with TCS have many characteristics that can trigger snoring, including nasal obstruction, mouth supplementary breathing, and retruded mandible with oropharyngeal impaired dimensions.[13] When there are no alterations in the sleep study, patients may benefit from sleep nasendoscopy and pharyngeal manometry, aiming for snoring diagnosis and treatment planning.[36]

Obstructive sleep apnea relates to impaired quality of life, like other chronic disorders such as arthritis, angina, hypertension, diabetes, and back problems.[37] Vitality and social functioning are the FS-36 domains exhibiting major mean differences – 24.0% and 27.9% – in individuals with OSA than the general population.[38] Vitality is defined as loss of energy or presence of fatigue, and changes of 12.5 points are used to determine a clinically significant threshold in this outcome.[8]

Previous evidence suggested that adults with TCS, especially females, have a low-status quality of life.[10] Detrimental correlations of the AHI with several domains of the SF-36 occurred in this study, suggesting that OSA affects several dimensions of a patient's life without sex predilection. Normative data for individuals from our geographical area, with a similar age range (25–34), and without OSA or craniofacial anomalies, determined an 81.3 vitality score.[39] Therefore, indicating a magnitude difference of 17.3%, compared with our findings, and evidencing a clinically significant modification of this parameter in adults with TCS.

In conclusion, adults with TCS are at risk of having OSA and this condition is associated with altered anthropometric measurements, increased systolic pressure, and impairment of some quality-of-life domains.

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