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Are home sleep studies useful in diagnosing obstructive sleep apnea in children with down syndrome?

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Abstract

Introduction and Aim: Obstructive sleep apnea syndrome (OSAS) is frequent in children with Down syndrome (DS) and polysomnography (PSG) is recommended for all children with DS. However PSG is not always available and alternative diagnostic methods are needed. The aim of the study was to evaluate the feasibility and validity of home polygraphy (HPG) in children with DS.

Methods: A national DS association was contacted and children aged 6 to 18 years who accepted to participate were recruited. Otorhinolaryngologic evaluation, in-lab PSG and HPG were performed. OSAS was diagnosed by PSG with an apnea-hypopnea index (AHI) more than or equal to 1. OSAS severity was classified as moderate-to-severe if AHI was more than or equal to 5. Receiver operating characteristic curves were calculated for HPG using PSG as the gold standard.

Results: Nineteen children (12 girls) completed the study. Median age was 11.3 years. Demographic and clinical characteristics were similar in children with and without OSAS. Eighty-nine percent of HPG studies were technically acceptable at the initial night and the success rate was 100% when two failed studies were repeated. PSG revealed OSAS in six (32%) children, two had mild and four had moderate-to-severe OSAS. All four patients with moderate-to-severe OSAS diagnosed with PSG have been diagnosed with the same severity on HPG. HPG had 100% sensitivity and 83% specificity when AHI ≥ 3 was set as diagnostic criteria.

Conclusion: HPG is a feasible and reliable test of OSAS in children with DS and may be useful in diagnosis and treatment of patients with moderate-to-severe OSAS in this patient group.

KEYWORDS

home sleep apnea test, polygraphy, polysomnography, sleep disordered breathing, sleep medicine

1 | INTRODUCTION

Down syndrome (DS) or trisomy 21 is the most common genetic disorder, occurring approximately in 1 of 900 live births.[1] Children with DS are at increased risk for development of obstructive sleep apnea syndrome (OSAS) compared to typically developing children. Phenotypic features of DS include midfacial and mandibular hypoplasia, small upper airway, relative macroglossia, posteriorly placed tongue, lymphoid hyperplasia, hypotonia, and obesity which may predispose to OSAS.[2] The prevalence of OSAS is reported to be between 24% and 95% in the DS population.[3-10]

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OSAS in children is associated with significant morbidity, such as growth failure, pulmonary hypertension, cor pulmonale, cognitive and behavioral problems, enuresis and decreased quality of life.[11] Children with DS may be at greater risk since they already have high incidence of cognitive, behavioral, and cardiologic problems.[12,13] Therefore prompt diagnosis and treatment of OSAS is essential. Although children with DS may present at very early ages with OSAS, American Academy of Pediatrics recommends referral to a pediatric sleep laboratory for a sleep study for all children with DS by 4 years of age due to the high prevalence of sleep disordered breathing in this population.[9,14]

Sensitivity and specificity of symptoms, sleep questionnaires and physical findings are inadequate in diagnosing OSAS. The gold standard for the diagnosis is overnight, attended, in-lab polysomnography (PSG) study.[11,15] However because of shortage of sleep laboratories with pediatric expertize, limited availability and expensiveness of this method, there is a need for alternative diagnostic methods.

Home sleep apnea testing with portable devices has been considered as an alternative as it is potentially more cost-effective, convenient, and accessible. The majority of studies report that it is feasible to perform home studies in children who are being investigated for OSAS, even in young pre-school children.[16-20] Currently the evidence on home testing with portable polygraphy devices in the diagnosis of OSAS in patients with DS is limited. The objective of the present study was to evaluate the feasibility and diagnostic validity of home polygraphy (HPG) in a community sample of children with Down syndrome by comparing it with in-lab PSG. We hypothesized that HPG would be feasible in children with Down syndrome and it would accurately identify the patients with OSAS.

2 | METHODS

2.1 | Study population

A national DS association was contacted, families were informed about OSAS and the terms of the study through the association's website. Families who were interested in the study were invited to participate. Children were eligible if they had a confirmed diagnosis of DS and if they were 6 to 18 years of age. Children who had unstable congenital heart diseases or children who had hypothyroidism or respiratory tract infection during the time of the study were excluded. The study was approved by the ethics committee of Istanbul University, Istanbul Medical Faculty (reference number 1133). Written informed consent was signed by the parents on behalf of their children.

2.2 | Patient characteristics

Age, sex, medical history including gestational age, snoring frequency, presence of observed apneas, history of adenoidectomy and/or tonsillectomy, parental history of smoking, snoring and adenoidectomy and/or tonsillectomy were noted. Height and weight were measured and body mass index (BMI) and body mass index *z*-score were calculated.[21] Children with a BMI *z*-score more than 2 were considered obese.

2.3 | Ear nose throat (ENT) examination

Examination of the upper airway was performed by an otorhinolaryngology specialist and palate position, size of the tonsils, adenoids, and lingual tonsils were evaluated. Palate position was graded according to Friedman palate position score.[22] Tonsil size was assessed according to Brodsky criteria.[23] Adenoid tissue size was graded from one to four by means of nasopharyngeal endoscopy according to Cassano classification.[24] Lingual tonsil size was scaled according to Friedman scale.[25]

2.4 | Sleep studies

Each patient underwent overnight, attended in-lab PSG in the Sleep Unit of Istanbul University, Istanbul Medical Faculty, Pulmonology Department. The recorded parameters were electroencephalography (C3A2, C4A1, O1A2, and O2A1), right and left oculography, chin electromyogram, anterior tibial electromyogram, electrocardiography, pulse oximetry and pulse waveform, thoracic and abdominal inductance plethysmography, nasal thermistor, nasal pressure transducer, audio recording and body position. Staging of sleep and the scoring of respiratory events and arousals were performed according to the American Academy of Sleep Medicine pediatric criteria.[26] Apnea-hypopnea index (AHI) was defined as the sum of all obstructive, central, and mixed apneas and hypopneas divided by hours of total sleep time. Hypopnea was defined as 30% reduction in airflow accompanied by a 3% decrease in oxygen saturation or an arousal from sleep. Oxygen desaturation index was defined as the number of times per sleep hour that oxygen saturation decreased more than or equal to 3%.

An unattended HPG was done at home within a week with a portable type3 device (NoxT3 portable sleep monitor; ResMed). Families were given instructions on how to place and remove the sensors and start and stop recording. Recorded parameters were nasal flow by means of a nasal flow cannula, chest, and abdominal movements by inductance plethysmography, heart rate, and oxygen saturation by pulse oximetry, snoring by microphone, and body position. AHI and oxygen desaturation index (ODI) were defined with the same criteria used in scoring PSG. OSAS was defined as an AHI more than or equal to 1 episode/h.[11]

After both sleep studies were performed parents completed a brief questionnaire rating the comfort and ease of application of both methods on Likert scale ranging from 1 to 5 (with 5 being the best) and preferred diagnostic modality was asked.

2.5 | Statistical analysis

Data were reported as mean ± SD for variables with normal distribution and median (25th-75th percentile) for variables which

were not normally distributed. Categorical variables were compared with the χ^2. The Mann-Whitney *U* test was used to compare continues variables of two independent groups. Spearman correlation test was used for correlation analysis of continuous variables. A $P < .05$ was considered as statistically significant. Bland-Altman graph was plotted to assess the agreement of PSG AHI and HPG AHI values. Receiver operating characteristic (ROC) curves were calculated for HPG using PSG as the gold standard.

3 | RESULTS

3.1 | Study population

A total of 39 families responded to the web announcement and were provided information about the study. Three children failed to meet the inclusion criteria: two of them did not fulfill the age criteria, one of them had unstable cardiac disease. Ten families decided not to participate due to the demands of the study like traveling to the study center and sleeping overnight in a sleep unit. Twenty-six children were recruited, five of them did not present on the day of their in-lab sleep study, two of them dropped out after PSG was performed and HPG was not done. The median age of the children who completed the study and who were not included in the study ($n = 20$) were 11.3 (10.3-14.3) and 9.5 (7.0-16.0), respectively and the difference was not statistically significant ($P = .39$). Twelve (63%) of the children who were evaluated were females, while most of the children who were not recruited or dropped out were male (80%) and gender profiles were statistically different ($P = .01$). Demographic and clinical characteristics of 19 children who completed the study are listed in Table 1. Habitual snoring in the absence of upper respiratory tract infection was reported in 10 (53%) and observed apnea during sleep was reported in 9 (47%) patients. The median BMI was 20.7 kg/m^2 (19.2-25.9), the median BMI *z*-score was 0.85 (−0.02 to 2.02) and 6 (32%) children were obese.

3.2 | ENT examination

Ten (53%) children had grades III or IV Friedman palate position score. Eight (42%) children had grade 3 or 4 tonsils, two (11%) had adenoid hypertrophy, eight (42%) had moderate/severe lingual tonsil hypertrophy.

3.3 | Home polygraphy

HPG was uninterpretable in two patients (11%) due to loss of airflow signal and had to be repeated. The findings of HPG of 19 children are summarized in Table 2. HPG revealed OSAS in 16 patients, 12 of them had mild and four of them had moderate/severe OSAS.

3.4 | In-lab polysomnography

PSG was successfully performed in all the children during the first night they attended the sleep laboratory. The results are summarized in Table 3. PSG revealed OSAS in six (32%) children, two of them had mild and four of them had moderate/severe OSAS.

TABLE 1 Demographic and clinical characteristics

Age (yrs), median (25-75 percentile)	11.3 (10.3-14.3)
Female sex *n*, (%)	12 (63)
BMI *z*-score, median (25-75 percentile)	0.85 (-0.02-2.02)
Obese *n*, (%)	6 (32)
Snoring *n*, (%)	10 (53)
Observed apneas during sleep *n*, (%)	9 (47)
Premature birth *n*, (%)	2 (11)
History of adenoidectomy *n*, (%)	4 (21)
History of tonsillectomy *n*, (%)	3 (16)
Maternal history of snoring *n*, (%)	8 (42)
Paternal history of snoring *n*, (%)	9 (47)
Indoor exposure to smoking *n*, (%)	8 (42)

Abbreviation: BMI, body mass index.

3.5 | Comparison of HPG and in-lab PSG

There was a significant correlation between the AHI and ODI obtained by PSG and by HPG ($r = 0.642$, $P = .003$ and $r = 0.629$, $P = .04$, respectively). However, HPG overestimated AHI and OAI compared with PSG. Using a cut-off value of AHI ≥ 1/h, HPG had sensitivity of 100%, specificity of 23% in determining OSAS with a positive predictive value of 38% and negative predictive value of 100%. All four patients with moderate-to-severe OSAS diagnosed with PSG (AHI ≥ 5/h) have been diagnosed with the same severity on HPG.

ROC curves were calculated for HPG AHI using PSG as the gold standard (Figure 1). For PSG-AHI ≥ 1/h, the best cut-off for HPG AHI was ≥3/h with a sensitivity of 100% and specificity of 85% in detecting OSAS in patients with DS. The area under the curve was 0.98 (Table 4). Bland-Altman graph was plotted to assess the agreement of PSG AHI and HPG AHI values (Figure 2).

HPG was found more comfortable and easy by the parents compared to in-lab PSG ($P = .025$) and 16 (84%) of the parents reported HPG as their preferred diagnostic modality.

TABLE 2 Home polygraphy findings

	Median (25th-75th percentile)
Total recording time (min)	499.0 (406.0-600.4)
Supine sleep time (% of total sleep time)	51.5 (29-77.5)
AHI (episodes/h)	2.2 (1.1-4.4)
Obstructive AHI (episodes/h)	2.2 (1.1-4.2)
OAI (episodes/h)	1.3 (0.3-1.1)
$SpO_{2\ mean}$ (%)	95.3 (94.2-96.9)
$SpO_{2\ nadir}$ (%)	80.0 (78.0-85.0)
ODI (episodes/h)	6.2 (2.8-17.9)

Abbreviations: AHI, apnea-hypopnea index; OAI, obstructive apnea index; ODI, oxygen desaturation index.

TABLE 3 PSG findings

	Median (25th-75th percentile)
Total recording time (min)	507.0 (478.5-529.0)
Total sleep time (min)	429.5 (376.0-462.0)
Stage N1 (% TST)	0.3 (0.2-0.5)
Stage N2 (% TST)	52.1 (44.8-65.1)
Stage N3 (% TST)	33.7 (24.8-44.9)
REM (% TST)	10.1 (6.6-14.0)
Supine sleep time (% of total sleep time)	56.5 (21.3-74.8)
AHI (episodes/h)	0.6 (0.3-2.2)
Obstructive AHI (episodes/h)	0.6 (0.2-1.6)
OAI (episodes/h)	0.3 (0.1-0.6)
$SpO_{2\ mean}$ (%)	96.6 (95.7-97.4)
$SpO_{2\ nadir}$ (%)	87.0 (82.0-91.0)
ODI (episodes/h)	3.6 (0.5-7)

Abbreviations: AHI, apnea-hypopnea index; OAI, obstructive apnea index; ODI, oxygen desaturation index; PSG, polysomnography; REM, Rapid eye movement sleep; TST, total sleep time.

4 | DISCUSSION

The present study suggests that HPG may be a feasible and a valid alternative to PSG in children with DS. Although overnight, attended, in-lab PSG is considered the gold standard test for the diagnosis of OSAS, other diagnostic modalities are recommended when in-lab PSG is not available.[11] Polygraphy has been shown to be feasible and has been used as alternative to in-lab PSG in typically developing children.[20] Current study showed that HPG was feasible with a high rate of technically acceptable studies in children with DS aged 6 to 18 years. Success rate was 89% at the initial night and 100% when repeated (two children who failed at the first night due to loss of airflow signal completed a second study). HPG was well tolerated by the children and families found it easier and more comfortable compared to in-lab PSG.

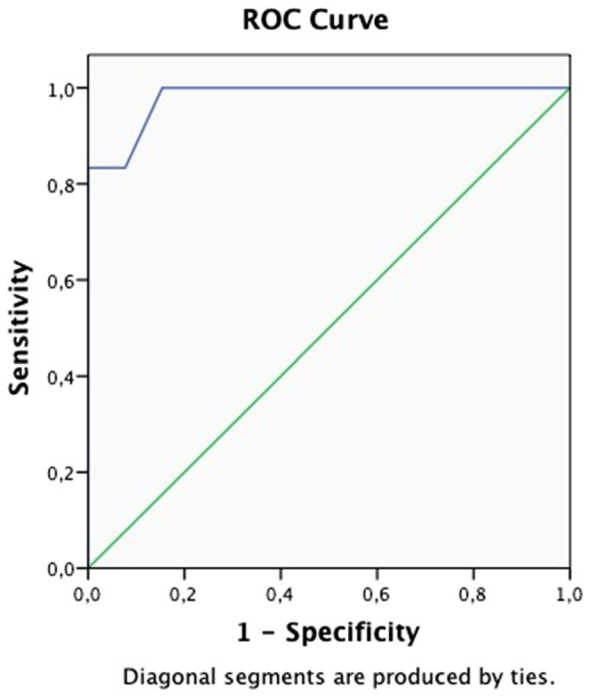

FIGURE 1 Receiver operating characteristic (ROC) curve for home polygraphy (HPG) apnea-hypopnea index (AHI) in determining obstructive sleep apnea syndrome (OSAS) with diagnostic criteria set as AHI ≥ 1 in polysomnography (PSG) [Color figure can be viewed at wileyonlinelibrary.com]

TABLE 4 Diagnostic validity indexes for HPG compared with in-lab PSG (PSG-AHI ≥ 1/h)

HPG AHI (episode/h)	Sensitivity (%)	Specificity (%)
≥1	100	30
≥1.6	100	47
≥2.1	100	61
≥3	100	85
≥3.9	83	93
≥4.3	83	100

Abbreviations: AHI , apnea hypopnea index; HPG, home polygraphy; PSG, polysomnography

Feasibility rates of HPG in previous home studies involving typically developing or former preterm children were similar to our study. Rosen et al[17] performed HPG in 850 children aged 8 to 11 years. Trained staff went to children's homes to apply the sensors and technically acceptable home PG studies were obtained in 94% of the children. Brockmann et al[18] recruited 101 children with median age 2.4 years and performed 75 HPG and 26 hospital-based PG.[18] The initial set-up of sensors was performed in the evening in the sleep lab by the staff and caregivers were taught how to check if the sensors were placed adequately and they were provided with a hotline number for the night. Technically acceptable and interpretable studies were obtained in 93% of patients and failure rates were similar in home and hospital setting.

To date there are few studies using polygraphy for diagnosing OSAS in children with DS. Recently in a study by Hill et al[6] HPG was performed in children with DS aged 0.5 to 5.9 years and 143 of 194 (74%) studies were successful at first attempt and 188 successful studies were achieved requiring an average of 1.3 studies. The lower rate of acceptable results compared with our study could be explained by the fact that the children recruited were younger children who may be less compliant. In a previous study assessing the prevalence of sleep-disordered breathing in children with DS, 108 children underwent polygraphy, however the study was performed at a hospital setting and the children were continuously observed by a trained technician.[5] Our study showed that high rate of technically acceptable recordings could be obtained in home setting in children with DS even in the absence of trained staff to visit the child's home and the set-up of HPG were performed by caregivers.

Alonso-Alvarez et al[19] performed a study to assess the diagnostic reliability of HPG in children with a clinical suspicion of OSAS and

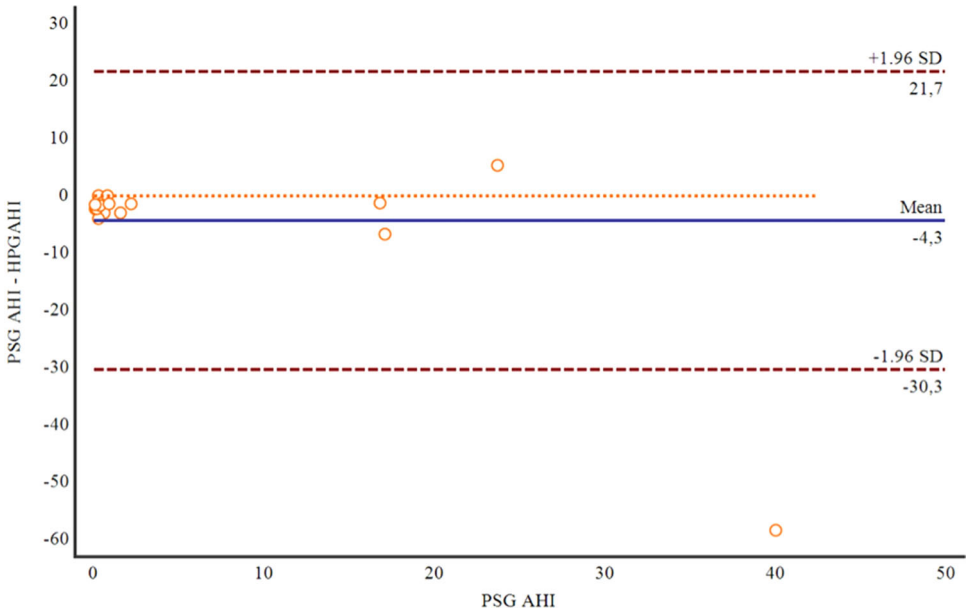

FIGURE 2 Bland-Altman graph was plotted to assess the agreement of polysomnography (PSG), apnea-hypopnea index (AHI), and home polygraphy (HPG) AHI values [Color figure can be viewed at wileyonlinelibrary.com]

50 children aged 2 to 14 years underwent an initial HPG followed by a later date, same night PSG and polygraphy in the sleep lab. Of the children 66% were diagnosed with OSAS based on a PSG-defined obstructive respiratory disturbance index (ORDI). No significant differences were found in the total number of apneas or hypopneas between the studies and HPG ORDI ≥ 5.6 exhibited a sensitivity of 90.9% (95% confidence interval [CI], 79.6%-100%) and a specificity of 94.12% (95% CI, 80%-100%). In a previous study by Rosen et al[17] a subgroup of 55 children in the study group had in-lab PSG and HPG performed within 3 months for comparison, and when using an AHI ≥ 5/h total sleep time (TST) as cut-off for the diagnosis of OSAS, the sensitivity of HPG was 88% and the specificity was 98%. Massicotte et al[27] studied 35 children with PSG and polygraphy simultaneously in the sleep lab and using a cut-off AHI ≥ 1.5 events/h, the manually scored polygraphy had a sensitivity of 94% and specificity of 16% to detect OSAS (AHI ≥ 1.5 events/h) diagnosed by PSG. When the cut-off was increased (AHI of ≥5 events/h) polygraphy still had good sensitivity of 94% and specificity increased to 61% in detecting OSAS (AHI ≥ 1.5 events/h) diagnosed by PSG.

Similar to these studies, the present study showed that HPG is sensitive in detecting OSAS also in DS. Although there are a few studies which polygraphy was used, to our knowledge this is the first study evaluating the validity and accuracy of HPG by comparing it with in-lab, attended PSG for diagnosis of OSAS in children with DS. The present study showed that AHI values of HPG and PSG are significantly correlated and HPG had excellent sensitivity and 85% specificity when AHI ≥ 3/h was used as a cut-off for the diagnosis of OSAS.

In the present study, we observed that HPG overestimated AHI compared with the PSG, similar to Massicotte et al[27] and incorrectly diagnosed patients with OSAS. The over-diagnosis of OSAS with polygraphy may be due to events scored during wakefulness, reduced flow observed on a nasal flow channel during mouth breathing even in the absence of apnea and/or post-arousal artifacts mistaken as reduced airflow.

The main limitation of this study was low number of the patients who were recruited. Currently in our country PSG is not routinely performed in children with DS due to shortage of pediatric sleep laboratories and most families are not aware of the consequences of OSAS. Few families responded to our announcement and in addition only half the patients who were contacted accepted to participate in the study because the families thought that the study was demanding or that their child would not be compliant. Another limitation was that we included patients older than 6 years of age. Although obstructive sleep apnea syndrome is observed in all ages in these children, we wanted to perform polygraphy studies at home without a technician as in real life and due to limited resources we included children older than 6 years of age to improve the compliance. Despite the low number of patients, this study has also strengths, we recruited our patients from the community and they had the gold standard test within a short period of time in their homes.

In conclusion, this study showed that HPG is a feasible and a valid test of OSAS in children with DS older than 6 years and may be useful in diagnosis and treatment of patients with moderate/severe OSAS in this patient group. However further studies involving more participants including younger children are needed to make definite statements regarding the feasibility and validity of HPG in children with DS.

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CONFLICT OF INTEREST

The authors declare that there are no conflict of interests.

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