Accuracy of Parental Perception of Nighttime Breathing in Children with Down Syndrome

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Abstract

Objective. In 2011, the American Academy of Pediatrics published a guideline for children with Down syndrome (DS), recommending a polysomnogram (PSG) by age 4 years regardless of symptoms. Their rationale was based on 2 publications with small cohorts, where at least 50% of the children had no obstructive sleep apnea (OSA) symptoms but their PSG results were abnormal. The American Academy of Otolaryngology—Head and Neck Surgery Foundation published a clinical practice guideline recommending PSG prior to adenotonsillectomy for these children. This study aimed to assess parents’ accuracy of their children’s breathing patterns as compared with PSGs in a larger cohort of children with DS.

Study Design. Case series with chart review.

Setting. Tertiary care academic pediatric hospital.

Subjects and Methods. Sleep intake forms assessing frequency of parent-observed apnea, snoring, and restless sleep were analyzed. None of the children had a previous tonsillectomy. Two groups were analyzed according to symptoms: infrequent (<3 nights per week on all questions answered) and frequent (≥6 nights per week on at least 1 question). OSA severity was categorized as follows: normal, <2 events per hour; mild, 2 to 4.9; moderate, 5 to 9.9; and severe, ≥10.

Results. A total of 113 children met inclusion criteria: 34% (n = 38) had infrequent symptoms, and 66% (n = 75) had frequent symptoms. Parents were unable to predict the presence or absence of OSA by nighttime symptoms (P = .60). The risk of OSA for children with frequent symptoms versus those with infrequent symptoms was 1.04 (95% CI, 0.89-1.3).

Conclusion. Parents of DS children are unable to predict the presence or absence of OSA by nighttime symptoms, nor are they able to determine its severity.

Keywords

Down syndrome, tonsillectomy, obstructive sleep apnea, child, polysomnography, history, symptoms

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Down syndrome (DS) is a common condition and is associated with congenital heart anomalies, pulmonary hypertension, and cognitive delays. Secondary to these children’s craniofacial structure and muscle tone, they are at increased risk for developing obstructive sleep apnea (OSA). Prevalence estimates of OSA in children with DS vary from 20% to 80%.1-3 The diagnosis and treatment of OSA are especially important for these children, since OSA has been implicated as a contributing factor to pulmonary hypertension,4 cardiovascular complications, failure to thrive, impaired cognition,5-7 and behavioral problems.8,9 Untreated OSA may exacerbate the preexisting conditions of these children.

In 2011, the American Academy of Pediatrics published clinical guidelines for children with DS recommending universal screening for OSA with a polysomnogram (PSG) by age 4 years, even in asymptomatic children.10 Their evidence to support this recommendation was based on 2 small case series. In a 5-year longitudinal study in which the otolaryngologic problems seen in DS were evaluated, Shott et al demonstrated that parental history was unable to predict the PSG findings.11 Fifty-four percent (13 of 24) had no

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OSA symptoms, yet their PSG results were abnormal, whereas 7 of 11 (64%) with OSA symptoms had normal PSG results. A smaller case series of 21 children also demonstrated that parental history was unable to accurately predict PSG findings. A recent study from Belgium further supports the American Academy of Pediatrics’ recommendation, since 54% of children that had no parent-reported OSA symptoms had an obstructive apnea-hypopnea index $\geq$2 events per hour. In 2011, the American Academy of Otolaryngology—Head and Neck Surgery also published a clinical guideline, and the authors made a strong recommendation to perform a PSG prior to a tonsillectomy for children with DS. The rationale was that, since physical examination and history are poor predictors of the presence and severity of OSA and children with DS have higher perioperative risks, one needs to be certain of the diagnosis prior to surgery. Also, those children with severe OSA may require additional preoperative evaluation and a higher level of inpatient monitoring. The objective of this study was to assess parents’ accuracy of their children’s breathing patterns in a larger cohort of children with DS who had not undergone a previous adenotonsillectomy, to determine if these clinical guidelines are reasonable and to assess for risk factors associated with an abnormal sleep study.

Methods

Participants

This study was approved by the Colorado Multiple Institutional Review Board. Children were identified retrospectively via an electronic medical record search that combined the diagnostic code for DS with the procedure code for polysomnography (Current Procedural Terminology: 95808, 95810, 95811; International Classification of Diseases, Ninth Revision: 89.17) and tonsillectomy (International Classification of Diseases, Ninth Revision: 28.2-28.3) from 2009 to 2015.

Charts of 208 children with DS who had a PSG and no prior tonsillectomy were reviewed. Children were included if their prestudy sleep intake form was completed. These forms are completed on the night of the sleep study, and they assess the frequency of parent-observed apnea, snoring, and restless sleep. Forms with answers to at least 2 of the 3 nighttime symptoms were retained for analysis. Two groups were analyzed according to symptoms: infrequent (<3 nights per week on all questions answered) or frequent (≥6 nights/week on at least 1 question). OSA severity was categorized as follows: normal, <2 events per hour; mild, 2 to 4.9; moderate, 5 to 9.9; and severe, ≥10.

Analysis Plan

All analyses were performed with GraphPad Prism 7.02 (GraphPad, San Diego, California). $P < .05$ was considered statistically significant. Fisher’s exact test was used to assess the relationship between symptom frequency and PSG results. A chi-square was used to evaluate the association between symptom frequency and OSA severity.

Results

Between 2009 and 2015, 208 children with DS but no prior tonsillectomy underwent PSG. Of those, 113 met the inclusion criteria. Fifty-three percent were girls, and the average age at preoperative PSG was 5.89 years. Forty-eight percent were Caucasian, 40% Hispanic, 4% African American, and 8% other. Significant comorbidities included congenital heart disease (71.75%), prematurity (27%), pulmonary hypertension (8.8%), and hypothyroidism (23%). Twenty-two percent were obese (body mass index for age ≥95%), and 20.4% were overweight (body mass index for age >85%).

Of the 113 who met inclusion criteria, 34% (n = 38) had infrequent symptoms and 66% frequent (n = 75). Significant comorbidities were not associated with either the presence of OSA or OSA severity. Parents were unable to predict the presence or absence of OSA by nighttime symptoms ($P = .60$). Table 1 summarizes the polysomnographic findings. The risk of OSA for children with frequent symptoms versus those with infrequent symptoms was 1.04 (95% CI, 0.89-1.3). As shown in Figure 1, the distribution of OSA severity did not differ between children with frequent and infrequent symptoms ($P = .85$). Parental report of sleep symptom frequency was not associated with significant comorbidities.

Discussion

OSA is a common condition with significant health risks for any child. Children with DS are at higher risk of OSA due to their craniofacial anatomy and muscle tone. To maximize the likelihood of there being a difference in parental accuracy in predicting OSA, the cohort was limited to children with DS whose parents reported either frequent or infrequent symptoms of disrupted sleep. Even so, our study demonstrated that parental accuracy in detecting OSA for this population is poor.

Regardless of the frequency of parent-reported OSA symptoms, our PSG results did not correlate with parents’ observations. Our results differed from Maris et al because they found that asymptomatic children were less likely to have OSA and, if OSA was present, it was less severe. Of note, the correlation of clinical assessment and presence of OSA by PSG is marginal even for otherwise healthy children. In the Childhood Adenotonsillectomy Trial, otolaryngologists assessed healthy children between the ages of 5 and 9 years by physical examination and reliance on parental history, and they could predict OSA only 50% of the time (ie, apnea-hypopnea index, >2 events per hour, or obstructive hypopnea index, >1 event/hour). For children with DS, the accuracy of parental report is worse. Besides the Maris et al study, smaller case series have demonstrated the high likelihood of having PSG-proven OSA even in the absence of symptoms. In 1991, Marcus et al evaluated a cohort of 53 children with either overnight or nap PSG. Although 68% of the children were asymptomatic, most of them still had an abnormal PSG results. Dyken et al evaluated a cohort of 19 children who were consecutively seen in a developmental DS clinic (8 of 19 had a previous adenotonsillectomy). Seventy-nine percent (15 of 19) had OSA by overnight PSG. Of those, 33%
There are several potential explanations for why parental observation did not correlate with PSG findings. First, the timing of the observation is important because REM sleep, when muscle tone is lowest, is preferential to the early morning hours. It is unknown when parents are observing their children’s breathing patterns. Second, parents were least likely to answer the question of whether apnea was present or absent. This may reflect a lack of knowledge of what apnea is, or the parents may have not observed their children’s sleep close enough to answer. Third, respiratory events can be subtle and not associated with significant increased work of breathing. While obstructive apnea has near-complete cessation of airflow with persistent respiratory effort, many respiratory events are partial pauses, or hypopneas, which are subtler.

It is unclear whether educating families for what to look for would improve parental accuracy. Although it is tempting to ask the parents to provide a video of the child’s characteristic breathing pattern, there may be a sampling error. A 1996 investigation that used a home videotape recording to diagnose OSA actually demonstrated great ability to detect OSA (sensitivity = 94%); however, there was an elevated number of false positives (low specificity), indicating that clinician review of videotapes actually overdiagnosed the condition. In the previous study, the parents did an excellent job capturing the children’s worst periods of sleep. However, the captured video may not have been representative of the entire night. A video alone has a selection bias and does not guarantee that the breathing pattern is typical for the entire night. The video can suggest intermittent peaceful breathing or even obstruction, but it does not allow one to fully characterize the severity of OSA, since it just a brief recording of the sleep.

Table 1. Polysomnographic Measures: Infrequent vs Frequent Symptoms.

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Infrequent (n = 38)</th>
<th>Frequent (n = 75)</th>
<th>P Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sleep architecture</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sleep efficiency, %</td>
<td>78.59 (15.16)</td>
<td>81.1 (10.33)</td>
<td>.302</td>
</tr>
<tr>
<td>Spontaneous arousal index</td>
<td>8.17 (5.66)</td>
<td>8.66 (6.31)</td>
<td>.6844</td>
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<tr>
<td>N1, %</td>
<td>8.24 (7.52)</td>
<td>9.72 (13.36)</td>
<td>.5271</td>
</tr>
<tr>
<td>N2, %</td>
<td>45.33 (17.04)</td>
<td>50.77 (19.87)</td>
<td>.1527</td>
</tr>
<tr>
<td>N3, %</td>
<td>30.93 (19.96)</td>
<td>23.3 (13.58)</td>
<td>.0182</td>
</tr>
<tr>
<td>REM, %</td>
<td>15.21 (9.81)</td>
<td>14.73 (8.10)</td>
<td>.7823</td>
</tr>
<tr>
<td>Respiratory events</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>OAH1</td>
<td>12.22 (11.26)</td>
<td>14.58 (14.97)</td>
<td>.3938</td>
</tr>
<tr>
<td>CAI</td>
<td>1.32 (1.20)</td>
<td>1.45 (1.87)</td>
<td>.7104</td>
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<tr>
<td>Oxygenation and ventilation</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mean wake O2, %</td>
<td>93.73 (3.06)</td>
<td>94.25 (2.33)</td>
<td>.3166</td>
</tr>
<tr>
<td>Mean sleep O2, %</td>
<td>92.82 (2.18)</td>
<td>93.17 (2.78)</td>
<td>.5049</td>
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<tr>
<td>Oxygen nadir, %</td>
<td>74.49 (7.32)</td>
<td>79.87 (6.68)</td>
<td>.785</td>
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<tr>
<td>O2 &lt;90%, % TST</td>
<td>16.61 (25.53)</td>
<td>14.16 (23.82)</td>
<td>.6151</td>
</tr>
<tr>
<td>Mean ETCO2, mm Hg</td>
<td>43.32 (3.64)</td>
<td>43.87 (3.71)</td>
<td>.4732</td>
</tr>
<tr>
<td>ET6C02 &gt;50, % TST</td>
<td>3.82 (10.52)</td>
<td>4.44 (12.03)</td>
<td>.7976</td>
</tr>
<tr>
<td>Limb movements: PLMI</td>
<td>1.75(4.53)</td>
<td>1.10 (2.70)</td>
<td>.343</td>
</tr>
</tbody>
</table>

Abbreviations: CAI, central apnea index; ETCO2, end-tidal carbon dioxide; OAH1, obstructive apnea-hypopnea index; PLMI, periodic limb movement index; TST, total sleep time.

*P < .05.

bMissing 1 data point.

cMissing 3 data points.

dMissing 5 data points.

Figure 1. Obstructive sleep apnea (OSA) severity by symptom frequency.

(5 of 15) with OSA had no report of snoring, and 75% (3 of 4) with no OSA did have snoring.

There are several potential explanations for why parental observation did not correlate with PSG findings. First, the timing of the observation is important because REM sleep, when muscle tone is lowest, is preferential to the early morning hours. It is unknown when parents are observing their children’s breathing patterns. Second, parents were least likely to answer the question of whether apnea was present or absent. This may reflect a lack of knowledge of what apnea is, or the parents may have not observed their children’s sleep close enough to answer. Third, respiratory events can be subtle and not associated with significant increased work of breathing. While obstructive apnea has near-complete cessation of airflow with persistent respiratory effort, many respiratory events are partial pauses, or hypopneas, which are subtler.

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Conclusion
Our data show that parental perception of sleep among children with DS does not accurately predict the presence or absence of OSA by nighttime symptoms in a large cohort. Children with DS, even with infrequent symptoms, are likely to have OSA. A parental report of no snoring, apnea, or restless sleep is a suboptimal method to decide that a sleep study is not necessary. In agreement with the clinical guidelines, our results support the recommendation of obtaining baseline PSGs in all children with DS by 4 years of age as well as prior to surgery, given the poor agreement between parent-reported child sleep symptoms and PSG-defined sleep apnea. Further prospective studies are needed to confirm and extend these results.

Author Contributions
Norman R. Friedman, study design and conduct, interpretation of data, writing and approval of manuscript; Amanda G. Ruiz, data collection, data analysis, editing and approval of manuscript; Dexiang Gao, data analysis, data interpretation, editing and approval of manuscript; David G. Ingram, study design and conduct, interpretation of data, writing and approval of manuscript.

Disclosures
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References