Screening for Obstructive Sleep Apnea in Children with Down Syndrome

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Objective To compare symptoms of obstructive sleep apnea (OSA) and polysomnography (PSG) results in children with Down syndrome and typically developing children.

Study design A total of 49 children with Down syndrome referred for PSG between 2008 and 2012 were matched with typically developing children of the same sex, age, and OSA severity who had undergone PSG in the same year. A parent completed a sleep symptom questionnaire for each child. Sleep quality and measures of gas exchange were compared between the matched groups.

Results The 98 children (46 females, 52 males) had mean age of 6.2 years (range, 0.3-16.9 years). Fourteen children had primary snoring, and 34 had OSA (9 mild, 7 moderate, and 19 severe). Children with Down syndrome had more severe OSA compared with 278 typically developing children referred in 2012. Symptom scores were not different between the matched groups. Those with Down syndrome had a higher average pCO_2 during sleep (P = .03) and worse McGill oximetry scores.

Conclusion Compared with closely matched typically developing children with OSA of comparable severity, children with Down syndrome had a similar symptom profile and slightly worse gas exchange. Referred children with Down syndrome had more severe OSA than referred typically developing children, suggesting a relative reluctance by parents or doctors to investigate symptoms of OSA in children with Down syndrome. These findings highlight the need for formal screening tools for OSA in children with Down syndrome to improve detection of the condition in this high-risk group. *(J Pediatr 2014;165:117-22)*.

Down syndrome, or trisomy 21, is the most common childhood genetic disorder, with an estimated incidence of 14.47 per 10 000 live births.[1] Children with Down syndrome are at increased risk for obstructive sleep apnea (OSA), with a reported prevalence of 31%-79%[2-5] compared with 1%-5% in the general pediatric population.[6] The characteristic facial features of Down syndrome—midfacial and mandibular hypoplasia, relative macroglossia, a shortened palate, and narrowed nasopharynx—are all anatomic risk factors for OSA, and hypotonia also may contribute to airway collapse during sleep.[7] Given the known behavioral and neurocognitive impacts of OSA in typically developing children,[8,9] the condition may be expected to have important implications for learning, development, and quality of life in children with Down syndrome. In addition, we have previously shown that the cardiovascular response to respiratory events (obstructive apnea and hypopnea) is blunted in children with Down syndrome, possibly increasing the hypoxia associated with OSA in these children.[10]

Given the high incidence and potential complications of OSA in children with Down syndrome, international guidelines have supported the use of routine polysomnography (PSG) for detection of OSA.[11,12] However, recent studies suggest that this practice is not widely implemented, with 65% of 249 children in one study never undergoing PSG.[13] Another study has suggested that parents of children with Down syndrome underestimate the significance of their child's sleep symptoms,[13] although objective measures of OSA severity using PSG were not included in that study. In the present study, we aimed to compare the parentally reported symptoms of OSA in children with Down syndrome referred for PSG with their PSG findings, and also to compare these findings with those in typically developing children. We hypothesized that parents of children with Down syndrome would accept noisy breathing during sleep as part of Down syndrome, and would report fewer symptoms and lesser impact on quality of life and daytime functioning for a given severity of OSA compared with parents of typically developing children. We also hypothesized that children with Down syndrome would have more severe OSA and worse gas exchange for a given severity of OSA.

AASM	American Academy of Sleep Medicine
BMI	Body mass index
OAHI	Obstructive apnea hypopnea index
OSA	Obstructive sleep apnea
PDSS	Pediatric Daytime Sleepiness Scale
PSG	Polysomnography
REM	Rapid eye movement
SpO_2	Oxygen saturation

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Methods

The database of the Melbourne Children's Sleep Centre was used to identify children with Down syndrome aged ≤18 years who underwent PSG for suspected OSA during the 5-year period from January 1, 2008, to December 31, 2012. Initially, 82 children were identified. Children who had undergone adenotonsillectomy (n = 18) or who also had another major developmental disability (n = 4; 3 with autism and 1 with Klinefelter syndrome) were excluded. Parents of 5 children did not provide consent for use of their child's data for research purposes. Four children had limited PSG data (<4 hours of sleep recorded) and thus were excluded from our analysis.

OSA was defined as an obstructive apnea hypopnea index (OAHI) of >1 event/hour. Severity of OSA was classified as follows: mild OSA, OAHI >1 and ≤5 events/hour; moderate OSA, >5 and ≤10 events/hour; severe OSA, >10 events/hour.

A cohort study was then performed by comparing the prevalence of OSA by severity category between the Down syndrome group and all children identified from the Melbourne Children's Sleep Centre database who had undergone PSG for suspected OSA in 2012 and had no significant comorbidity (n = 278). A case-control study was also performed by choosing controls from the Sleep Centre database for comparison with the Down syndrome group, matched for sex, age (closest possible, within 2 years), OSA severity, and PSG performed in the same year. Owing to the large variations in OAHI in the children with severe OSA (ie, OAHI >10 events/hour), children in this group were matched more closely, using the closest possible OAHI in age-matched control children (median difference, 3.7 events/hour; range, 0.3-13.9 events/hour). A sufficiently close match could not be found for 2 children with Down syndrome (1 with an OAHI of 135 events/hour), leaving 49 children with Down syndrome and 49 matched controls.

For each child, a parent completed the OSA-18[14] and Pediatric Daytime Sleepiness Scale (PDSS)[15] questionnaires on the night of the PSG study. The OSA-18 questionnaire is a disease-specific quality of life tool comprising 18 items in 5 domains as reported by parents: sleep disturbance, physical suffering, emotional distress, daytime problems, and caregiver concerns. A score of <60 suggests a small impact on health-related quality of life, a score of 60-80 suggests a moderate impact, and a score >80 suggests a large impact.[14] The PDSS questionnaire evaluates symptoms of daytime sleepiness using 8 items, each scored between 0 and 4. The mean total score in the original study was 15.3 ± 6.2 (range, 0-32), with higher scores associated with less sleep time and worse school-related outcomes and mood.[15] Although these questionnaires were validated in typically developing children, they were used in the present study as measures of symptom severity as perceived by parents.

Ethical approval for this study was granted by the Monash Health and Monash University Human Research Ethics Committees.

PSG Studies

Each child underwent a full overnight attended PSG in a pediatric sleep laboratory using a commercially available sleep system (Compumedics, Melbourne, Australia). Height and weight were recorded at the time of the PSG and used to calculate body mass index (BMI).

Electroencephalography (C3/A2, C4/A1, O1/A2, and O2/A1), electrooculography, submental electromyography, electrocardiography, and left and right leg electromyography and body position were recorded. Oxygen saturation (SpO_2) was measured by pulse oximetry using Masimo RadicalSET (Irvine, California) or Bitmos (Dusseldorf, Germany) oximeters, both of which use Masimo signal extraction technology for signal processing and were set at a 2-second averaging time. Thoracic and abdominal breathing movements were recorded by uncalibrated respiratory inductance plethysmography (Pro-Tech zRIP effort sensor; Pro-Tech Services, Mukilteo, Washington). Transcutaneous carbon dioxide was measured using a TCM4/40 or TINA TCM3 monitoring system (Radiometer, Copenhagen, Denmark). Airflow was measured by nasal pressure (Salter-Style; Salter Labs, Arvin, California) and oronasal airflow (Sandman BreathSensor, Child Airflow Thermistor; Tyco Healthcare, Gosport, United Kingdom).

Scoring of PSG studies was conducted by trained technicians, who maintained a concordance rate >85% for both sleep and respiratory events. PSGs were performed during a period when the sleep laboratory moved from using American Thoracic Society criteria[16] to using the 2007 American Academy of Sleep Medicine (AASM) scoring criteria.[17] The key difference between the old and new scoring systems is the requirement for a 50% fall in airflow for a hypopnea under the AASM rules, compared with the 20% (a "discernible decrease") used previously. For this reason, the OAHI used for all studies included events with a 20%-50% drop in airflow, termed hypopneas, from 2008 to 2011 and respiratory event–related arousals in 2011 and 2012. Thus, obstructive apnea was defined as a decrease in flow signal to <10% of baseline in the presence of continued or increased respiratory effort. Obstructive hypopnea was defined as a reduction in flow signal of at least 20% from baseline in the presence of respiratory effort (with paradox or phase shift), associated with snoring or noisy breathing at event termination in conjunction with an arousal, awakening, or ≥3% SpO_2 desaturation.

Sleep was staged as non–rapid eye movement sleep stages 1, 2, 3, and 4 and rapid eye movement (REM) sleep from 2008 to 2011, with non–REM 3 and 4 combined into total slow-wave sleep for consistency with the AASM criteria adopted in 2012. A desaturation index ≥4% was defined as the number of times per hour that the SpO_2 dropped by 4% or more when associated with a central or obstructive respiratory event. Severity of abnormality on oximetry was