How helpful are sleep studies in determining surgical need in infants with achondroplasia?

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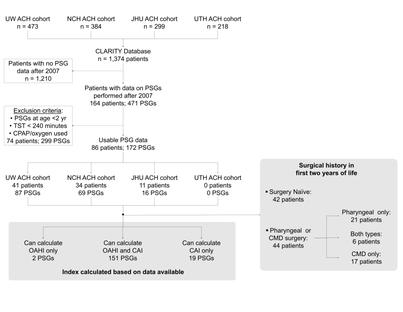
INTRODUCTION

- Achondroplasia is the most common form of dwarfism and disproportionate short stature with an incidence of 1:25K-30K. The majority of patients are born to average stature parents.
- Infants with achondroplasia are known to have an increased risk for sudden death due to critical craniocervical junction stenosis (CCJS).
- Neuroimaging and infant polysomnography (PSG) are recommended after diagnosis to help detect CCJS and decrease the risk of sudden death.
- We sought to clarify the natural history of PSG indices over the first 2 years in infants with achondroplasia with and without surgery (adenoidectomy (AD) and cervicomedullary decompression (CMD)) and compare indices after surgery to assess utility of interventions. Knowing baseline trends and indices helps interpret the PSGs.

METHODS:

- Retrospective data were abstracted from multisite CLARITY ACH data base from years 2008-2017. Centers include University of Wisconsin, University of Texas Houston, Johns Hopkins University, and Nemours Childrens Hospital Delaware.
- We evaluated PSG data including Obstructive Apnea Hypopnea Index (OAHI/OI), Central Apnea Index (CAI/CI), Total sleep time (TST,) AD surgery, CMD surgery, age. Only patients with adequate information, TST>240 minutes, and PSG performed on room air (RA) were included.
- We compared patients who were surgically naïve (SN) without AD or CMD, patients who only underwent AD, and patients who only underwent CMD.
- Statistical analyses include linear mixed models, Kuskal-Wallis test, and Fisher exact test.



RESULTS

172 PSGs in 86 patients with sufficient data. 153 PSGs from 80 patients were used to calculate OAHI and 170 PSGs from 85 patients used to calculate CAI. 151 PSGs in 80 patients used to calculate both OAHI and CAI. 23 patients underwent CMD and 27 patients underwent pharyngeal surgery. First PSG was obtained at a median of 0.58 years in SN patients and 0.50 years in patients who underwent surgery.

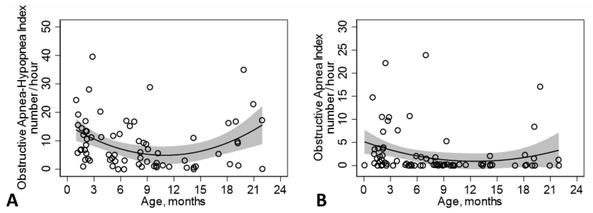
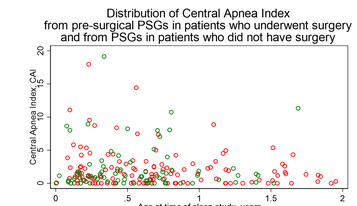


Figure 1: 2 yr temporal trends for (**A**) OAHI, (**B**) OAI, © HI, (**D**) CAI



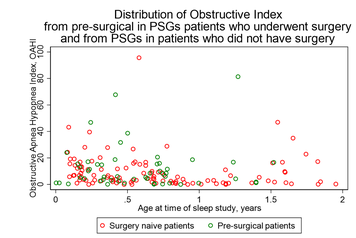


Figure 2: Distribution of CAI and OAHI in SN patients vs patients who went on to AD or CMD

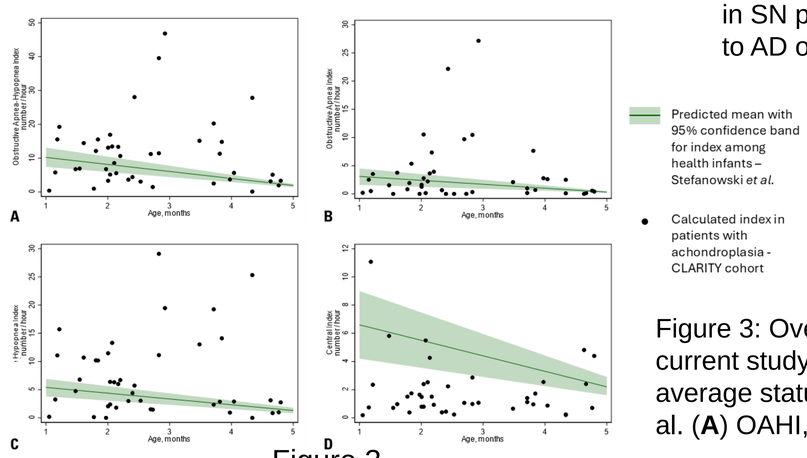


Figure 3: Overlay of PSG patient indices from current study and 95% CI bands in green from average stature children from Stefanovski et al. (**A**) OAHI, (**B**) OAI, (**C**) HI, (**D**) CAI

Figure 3

Procedure	Change OAHI (95% CI)	Change CAI (95% CI)	Median time post procedure
Adenoidectomy	**-14.1 (-23.5, -4.7)**	0.2 (-1.0, 1.5)	4.8 months
Cervical medulary decompresion	1.2 (-10.3, 12.8)	**-3.3 (-5.1, -1.5)**	5.7 months

CONCLUSIONS

- This is the largest sample of infants with achondroplasia and sleep studies in the literature.
- In surgically naïve patient/s with achondroplasia, PSG indices tended to improve over time until 2 years when obstructive events increase.
- Sleep studies were not predictive of who went on to require surgery. There were no significant differences in baseline OAHI or CAI indices or age of PSG between those who were SN or those who underwent surgery suggesting other important clinical factors play into decision making.
- Infants with achondroplasia had much larger variability in indices when compared to average stature infants, possibly contributing to the lack of futility in sleep studies alone to determine the need for surgery
- Surgery altered the trajectory of PSG indices:
  AD ➔ statistically improved OAHI
  CMD ➔statistically improved CAI

Limitations: This is a retrospective study. Many children did not have follow up sleep studies if they were clinically improved given the difficulty in obtaining them in young children. In those patients who had post-op sleep studies, they were not obtained at a specific interval.

Further studies: Prospective studies in children with achondroplasia with detailed and specific intervals may potentially help define parameters on PSG that suggest the need for AD or CMD and the improvement after these interventions..

ACKNOWLEDGEMENTS

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