

Obstructive sleep apnea in children with Beckwith-Wiedemann syndrome (2019)

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Background

Macroglossia is the most common feature in patients with Beckwith-Wiedemann syndrome (BWS). Patients with craniofacial abnormalities are known to have an increased risk for obstructive sleep apnea (OSA), or disordered sleep due to airway blockage. The prevalence and risk factors of OSA in patients with BWS and macroglossia is unknown.

Purpose

This study evaluated the prevalence of OSA in patients with BWS treated at Children's Hospital of Philadelphia (CHOP). All patients underwent a complete evaluation by genetics, plastic surgery, and pulmonology, and all had a sleep study (polysomnogram).

Findings

This study included 26 patients with BWS and macroglossia. The most common molecular subtype was loss of methylation at imprinting control region 2 (IC2 LOM), however patients with other BWS molecular subtypes were also evaluated. The degree of OSA did not differ between patients with IC2 LOM and other subtypes. The majority of patients had a history of snoring and/or noisy breathing during sleep prior to having a sleep study. The degree of OSA was significantly worse in younger patients, particularly those less than 6 months old. A significant relationship between OSA severity and BWS feature severity was found, as patients with higher BWS clinical score tended to have higher obstructive apnea-hypopnea indexes.

Following the sleep study, 9 patients underwent tongue reduction surgery and 1 patient had persistent OSA that was successfully treated with continuous positive airway pressure (CPAP). More research is needed to understand the benefits of a tongue reduction surgery as treatment for OSA in patients with BWS.

Conclusion

Patients with BWS and macroglossia have a high prevalence of OSA. It is important to evaluate for OSA in these patients, particularly those at younger ages and those presenting with more features of BWS.

Key Points

- Patients with BWS and macroglossia are at an increased risk for developing OSA.
- OSA tends to be most severe in patients under 6 months of age and those with more severe BWS features.
- Patients with BWS and macroglossia should be evaluated by specialists in genetics, plastic surgery, and pulmonology and receive a sleep study to determine the severity of OSA and best treatment plan.

Reference

Cielo CM et al. Obstructive sleep apnea in children with Beckwith-Wiedemann syndrome. *Journal of Clinical Sleep Medicine*. 2019;15(3): 375-381. PubMed PMID: 30853040