Role of Non-invasive ventilation in the management of obstructive sleep apnea in children with Beckwith-Wiedemann syndrome: Our experience

* NNagarjun¹, Bharath Reddy¹, and Kavya C¹

¹Bangalore Hospital

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Clinical Correspondence

Role of Non-invasive ventilation in the management of obstructive sleep apnea in children with Beckwith-Wiedemann syndrome: Our experience

Authors: K.R. Bharath Kumar Reddy. N. Nagarjun , Kavya C

Department of Pediatric Pulmonology and Sleep, Shishuka Childrens Specialty Hospital, Bangalore, Karnataka, India.

Authors:

1. Dr Bharath Kumar Reddy: MD (Paed), Consultant Pediatric Pulmonologist and Sleep Consultant
2. Dr N. Nagarjun : MD (Paed)
3. Dr Kavya C: MD (Paed)

Corresponding author:
Dr N.Nagarjun
Email ID: drnagarjun.n@gmail.com

To the editor,

Beckwith-Wiedemann syndrome (BWS) is a complex genetic disorder characterized predominantly by macroglossia, hemihyperplasia, and omphalocele. (1) Sleep disordered breathing is highly prevalent (nearly 48%) in these children due to the macroglossia and facial hemihypertrophy. (2) While surgical interventions such as glossectomy have been commonly employed to manage obstructive sleep apnea (OSA) in BWS, there has been limited experience or published data on the efficacy of conservative management with only non-invasive ventilation (NIV) in these children. (1) We present our experience of a child with BWS who demonstrated a significant response to NIV therapy.

Our patient was a 4-year-old male child, born to non-consanguineous parents, with classical features of BWS, including macroglossia and facial hemihypertrophy. Genetic testing confirmed the diagnosis of BWS with an imprinting abnormality on the IC2(KvDMR) domain of chromosome 11p15. The child was referred to us for complaints of snoring, gasping, and apneic episodes noted by parents during sleep, associated with excessive daytime sleepiness and irritability. Polysomnography conducted revealed severe OSA with an apnea-hypopnea index (AHI) of 10.5/hour associated with significant desaturations during the events. A drug-induced sleep endoscopy (DISE) revealed obstruction at the level of the tongue base with no significant adenoidal enlargement. Parents, when given both surgical and NIV options, preferred to avoid surgery and...
thereafter the child was initiated on continuous positive airway pressure (CPAP) therapy in a hospital-based protocol. On follow-up over the next three months, parents noted a significant improvement in sleep symptoms with no snoring or apneic episodes. A titration study was conducted and pressures were set at 7 cm of H20. The child was followed up on a monthly basis with repeated counseling on NIV adherence and troubleshooting when needed. The child remained compliant and satisfied with therapy over one-year, showing no recurrence of sleep symptoms. A repeat polysomnography conducted showed an AHI of <1/hour on a CPAP pressure of 7 cm of H20.

OSA is a common feature in children with BWS, often attributed to macroglossia and craniofacial abnormalities.(1) While surgical interventions like glossectomy have been traditionally considered, our case demonstrates the effectiveness of NIV alone in managing OSA associated with BWS. Surgical interventions reducing the tongue size, although effective in some cases in alleviating airway obstruction,(3) may not be the preferred option by most parents. Further, glossectomy being an invasive procedure can be associated with risks such as bleeding, infection, and post-operative pain. Additionally, long-term outcomes regarding speech function and taste perception remain a concern, as some studies have reported persistent tongue abnormalities and taste deficits following surgery. (2,4,5)

Conservative management with NIV offers a non-invasive alternative for managing OSA in BWS. CPAP therapy, if initiated in a child friendly hospital based protocol, followed by physician led strategies to ensure compliance, can play a significant role overcoming sleep symptoms and improving the quality of life in children with BWS. In the future, if more sleep centers opt for NIV therapy over surgical management in children with BWS, adequate comparative studies and protocols can be published to assist parents and physicians to make an evidence based decision.

References