CASE REPORT

Neurally Adjusted Ventilatory Assist: An Early Clue to Diagnosis of Congenital Central Hypoventilation Syndrome

Abdul Rauf[1], Dhiren Gupta[2], Anil Sachdev[3], Neeraj Gupta[4], Suresh Gupta[5], Praveen Kumar[6], Ramakant Sabharwal[7]

Abstract

Congenital central hypoventilation syndrome (CCHS) is characterized by shallow breathing during sleep due to negligible ventilatory sensitivity to hypercarbia and hypoxemia. It is diagnosed using a genetic test for PHOX2B mutation, which is not easily available. Neurally adjusted ventilatory assist (NAVA) is a spontaneous ventilatory mode that was designed basically for better adapting the ventilator to the patient by using electrical activity of diaphragm (EAdi) signals. We report a case of a 6-month-old infant who presented with recurrent apneas, where differential decrease in EAdi discharges during sleep using NAVA served as an early clue to the diagnosis of CCHS. Definitive diagnosis was later confirmed by genetic testing.

Keywords: Congenital central hypoventilation, Electrical activity of diaphragm, Neurally adjusted ventilatory assist.

Indian Journal of Critical Care Medicine (2019): 10.5005/jp-journals-10071-23286

Background

Congenital central hypoventilation syndrome (CCHS) is a disease characterized by shallow breathing during sleep due to negligible ventilatory sensitivity to hypercarbia and hypoxemia.[1] The diagnosis of this disease is confirmed by a genetic test for PHOX2B mutation, which is not easily available. Neurally adjusted ventilatory assist (NAVA) is a newer ventilator mode that uses electrical activity of diaphragm (EAdi) signals to detect neural respiratory drive and proportionally support the respiratory efforts. Neurally adjusted ventilatory assist helps improve patient ventilator interaction and decrease asynchrony during spontaneous breathing on the ventilator.[2]

We report here a distinctive use of the NAVA mode for early diagnosis of CCHS in an infant presenting with recurrent apneas. In our case, the differential decrease in EAdi discharges during sleep using NAVA served as an early clue to the diagnosis of CCHS and definitive diagnosis was later confirmed by genetic testing.

Case Description

A 6-month-old infant was admitted with acute history of respiratory distress following a viral prodrome. The child was initially managed on nasal prong oxygen. On day 2 of hospital stay, he developed an episode of apnea followed by desaturation and bradycardia. The child was intubated and mechanically ventilated. On reviewing the history, it was revealed that the child was born at full-term gestation and had an uneventful perinatal course for first 3 days after birth. On day 4 of life, the child was admitted in neonatal intensive care unit (NICU) for concern of recurrent apneas. Investigations including sepsis screen, neurosonogram, and cerebrospinal fluid (CSF) analysis were normal. He was managed conservatively and discharged after 10 days. Subsequently, he had two episodes of apnea at 3 and 4 months of life, both episodes occurred following persistent crying. These episodes were managed as probable breath-holding spells outside. During the current hospital course, child had recurrence of multiple episodes of apnea on the ventilator, when put on the spontaneous mode continuous positive airway pressure (CPAP). The child developed hypotension requiring inotrope on day 2 of hospital stay. But on day 3, he had severe hypertension needing intravenous antihypertensive infusion, suggesting autonomic instability. In the etiological workup, serum electrolytes and hemoglobin were in the normal range, sepsis screen was negative, EEG was normal, and baseline metabolic workup [arterial blood gas (ABG), serum ammonia, and lactate] and neuroimaging (MRI brain) were also normal. Echocardiography showed right ventricular hypertrophy with moderate pulmonary arterial hypertension. Clinical history and echo findings suggested a chronic etiology. We changed the ventilatory mode from the pressure control mode to the NAVA mode to monitor neural respiratory drive using EAdi. The EAdi signals were present during the awake state and absent or low during sleep, as shown in Figure 1. This indicated that respiratory drive was poor during the sleep. A normal nerve conduction velocity (NCV) test of phrenic nerves ruled out neuropathy as a cause of the poor EAdi signal. Possibility of central hypoventilation syndrome was considered and genetic testing was done. The PHOX2B gene sequencing report was available after 1 week; heterozygous in-frame duplication in the 20 alanine stretch of PHOX2B gene indicated common polyalanine repeat expansion mutation (PARM), which confirmed the diagnosis of CCHS. After counseling the parents, the child was discharged on home ventilation with bilevel positive airway pressure (BiPAP). The option for a diaphragm pacemaker was

[1–5]Department of Pediatrics (Intensive Care), Sir Ganga Ram Hospital, New Delhi, India

[6,7]Department of Pediatrics (Neurology), Sir Ganga Ram Hospital, New Delhi, India

Corresponding Author: Dhiren Gupta, Department of Pediatrics, (Intensive Care), Sir Ganga Ram Hospital, New Delhi, India, Phone: +91 9910455753, e-mail: dhireengupta@gmail.com

How to cite this article: Rauf A, Gupta D, Sachdev A, Gupta N, Gupta S, Kumar P, *et al.* Neurally Adjusted Ventilatory Assist: An Early Clue to Diagnosis of Congenital Central Hypoventilation Syndrome. IJCCM 2019;23(11):536–537.

Source of support: Nil

Conflict of interest: None

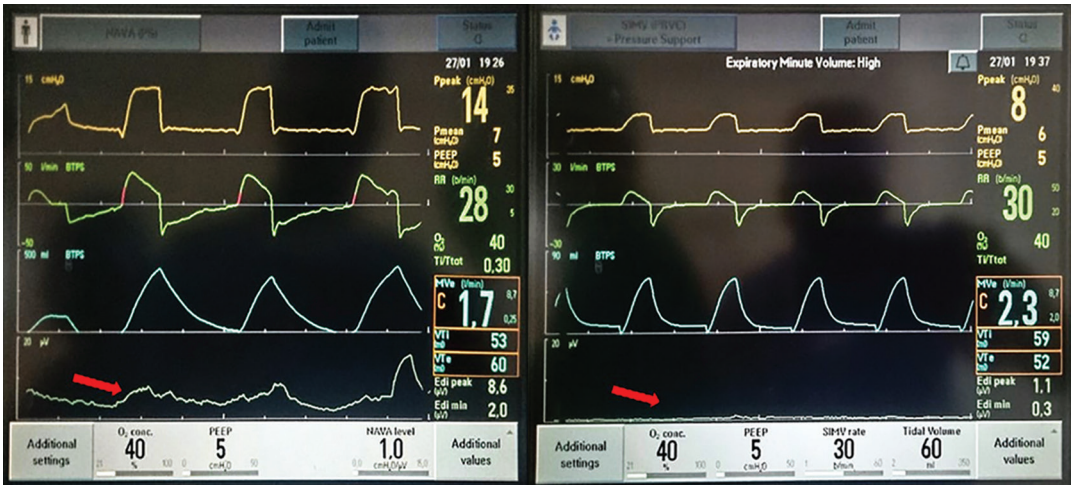

Fig. 1: Ventilator monitor showing adequate electrical activity of diaphragm discharges (pointed by arrows) in the neurally adjusted ventilatory assist mode during the awake state (left) and absent EAdi discharges (flat EAdi graph, pointed by arrows) during sleep (right). Note that the patient required the mandatory mode synchronized intermittent mandatory ventilation (SIMV) during sleep due to absence of spontaneous breaths

considered and a plan was made to procure and implant a diaphragm pacemaker once the child crosses infancy.

Discussion

Apnea occurs usually during sleep in CCHS; however, awake hypoventilation and breath-holding spells as seen in our case can also be the presenting features.[3] Autonomic instability is another salient clinical finding described in CCHS, which was present in our case.

Neurally adjusted ventilatory assist is a relatively new spontaneous ventilatory mode designed basically for adapting ventilatory support to the patient's actual demand by using EAdi signals. Rhythmic discharges from the respiratory centers in the brain pass through the phrenic nerves and stimulate the diaphragm to cause contraction. The EAdi signals are recorded by a special catheter (Edi catheter), which is inserted like a nasogastric tube and positioned at the crural part of the diaphragm. Here, the patient regulates the frequency and tidal volumes through his neural drive providing better interaction with the ventilator. Absent or decreased EAdi signals can occur due to decreased respiratory center activity or due to neuropathy involving phrenic nerves. Pediatric studies have shown that NAVA is a safe mode that can decrease asynchrony thus providing better comfort and lower sedation requirement.[4]

Diagnosis of CCHS is to be considered in a child with hypoventilation and hypoxemia without evident metabolic, infective, cardiac, neuromuscular diseases or an identifiable brainstem lesion.[5] Definitive diagnosis is established using the genetic testing for PHOX2B mutation, which is not easily available in developing countries, and results of these tests are also delayed in the available centers. In this context, the NAVA mode can be a useful adjunct for early diagnosis. There are very few reports in literature where the NAVA mode was used for this purpose and ours is the first report from India. Rahmani et al. demonstrated the absence of the EAdi signal in the quiet phase of sleep in a newborn baby suspected to have CCHS.[6] Recently, Sinclair et al. also reported a case where they captured central apnea during sleep using NAVA technology in a newborn child, who was later confirmed to have CCHS.[7] In our case also, the NAVA mode demonstrated decreased respiratory drive during sleep and gave us an early clue to the diagnosis. We feel that, in a child presenting with recurrent apneas where all common causes have been ruled out, NAVA may be used as an adjunct for diagnosing CCHS.

Conclusion

The differential decrease in neural respiratory drive during sleep, demonstrated by the NAVA mode, can serve as an early clue to the diagnosis of CCHS.

References

1. Chen ML, Keens TG. Congenital central hypoventilation syndrome: not just another rare disorder. Paediatr Respir Rev 2004;5(3):182–189. DOI: 10.1016/j.prrv.2004.04.009.
2. Beck J, Emeriuad G, Liu Y, Sinderby C. Neurally-adjusted ventilator assist (NAVA) in children: a systematic review. Minerva Anestesiol 2016;82(8):874–883.
3. Muzumdar H, Arens R. Central alveolar hypoventilation syndromes. Sleep Med Clin 2008;3(4):601–615. DOI: 10.1016/j.jsmc.2008.08.006.
4. Andrade LB, Ghedini RG, Dias AS, Piva JP. Neurally adjusted ventilatory assist in pediatrics: why, when, and how? Rev Bras Ter Intensiva 2017;29(4):408–413. DOI: 10.5935/0103-507X.20170064.
5. American Thoracic Society. Idiopathic congenital central hypoventilation syndrome: diagnosis and management. Am J Respir Crit Care Med 1999;160(1):368–373. DOI: 10.1164/ajrccm.160.1.16010.
6. Rahmani A, Rehman NU, Chedid F. Neurally adjusted ventilatory assist (NAVA) mode as an adjunct diagnostic tool in congenital central hypoventilation syndrome. J Coll Physicians Surg Pak 2013;23(2):54–56. DOI: 02.2013/JCPSP.154156.
7. Sinclair R, Teng AY, Jonas C, Schindler TM. Congenital central hypoventilation syndrome: a pictorial demonstration of absent electrical diaphragmatic activity using non-invasive neurally adjusted ventilatory assist. J Paediatr Child Health 2018;54(2):200–202. DOI: 10.1111/jpc.13690.