CASE REPORT

Neurally Adjusted Ventilatory Assist (NAVA) Mode as an Adjunct Diagnostic Tool in Congenital Central Hypoventilation Syndrome

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ABSTRACT

A full term female newborn was admitted to the neonatal intensive care unit (NICU) for continuous observation of apnea. Infant was noted to have apnea while asleep requiring intubation and mechanical ventilation. A video EEG was performed which demonstrated normal awake background without any seizure activity. Neurally adjusted ventilatory assist (NAVA) demonstrated the absence of electrical activity of the diaphragm (Edi) when the patient was in quiet phase of sleep. This finding on NAVA monitor raised the suspicion of central hypoventilation syndrome (CCHS) which was confirmed by genetic identification of the PHOX2B mutation.

Key words: *Neurally adjusted ventilatory assist (NAVA). Central apnea. Congenital hypoventilation syndrome. PHOX2B gene.*

INTRODUCTION

Neurally adjusted ventilatory assist (NAVA) is an innovative mode of ventilation helping the patient to control and augment their breathing as well as ventilator.[1] A special feeding catheter designed for NAVA (Edi catheter) is advanced through esophagus, and positioned at the crural part of the diaphragm to detect the diaphragmatic electromyogram (EMG).

The normal act of breathing depends on rhythmic discharges from the respiratory centre of the brain. The discharge travels along the phrenic nerve, and stimulates the diaphragm muscle cells, causing the muscles to contract. The diaphragm dome descends, which causes a drop in airway pressure and the inflow of air to enter the lungs. With NAVA, the electrical activity of the diaphragm (Edi) is captured using an electrode mounted to an Edi catheter, fed to the ventilator, and used to assist the patient's breathing. NAVA cycles are switched as soon as neural inspiration starts.

New neonatal ventilation methods have been developed recently to utilize new technological tools to detect the respiratory signal at a more proximal level in the respiratory cycle for an improved patient ventilator synchrony.[2,3]

NAVA offers clinical and diagnostic innovations as the ventilator trigger, works in concert with the diaphragm, encouraging its use, and minimizing its atrophy. It can also be a valuable tool in identifying true central apnea.[1]

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Central sleep apnea (CSA) is a sleep-related disorder in which the effort to breath is diminished or absent, typically for 10 – 30 seconds, either intermittently or in cycles and is usually associated with a reduction in blood oxygen saturation.[4,5] CSA is usually due to instability in the body's feedback mechanisms that control respiration.[4]

We report a case of central sleep apnea in a full term neonate diagnosed with the assistance of NAVA technology which demonstrated absence of diaphragmatic activity during the stage of quiet sleep by simultaneous EEG and diaphragmatic EMG activity recording.

CASE REPORT

A term female newborn was delivered with Apgar score of 9, 10 and 10 via cesarean section to a third gravida with obstetric history. Pregnancy was complicated by polyhydramnios of unknown etiology, which was treated with amniocentesis, otherwise pregnancy had progressed normally.

The baby needed initial steps of resuscitation and received Naloxone due to apneic spells in between breathing at birth as the mother had received Pethidine 4 hours prior to delivery of this baby. Infant birth measurements were within normal limits (weight 3030 gm, length 49.5 cm, head circumference 37.5 cm).

Infant was admitted to the Neonatal ICU for continuous observation of apnea following Naloxone administration in the delivery room. Apnea was noted to recur every time she slept, severe enough to require intubation and mechanical ventilation. Extensive metabolic work up as well as neuroimaging failed to reveal any cause for apnea. Detailed neurological examination followed by video EEG was performed which demonstrated normal awake background without any seizure activity.

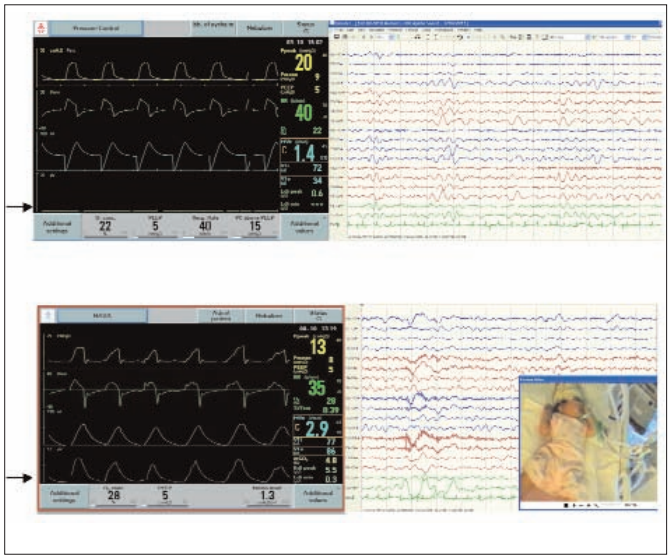

Figure 1: NAVA screen showing no Edi signals and video EEG (in sleep) above and with Edi signals with video EEG below (awake).

Neurally adjusted ventilatory assist was used as a mode of ventilation and was found to have absence of diaphragmatic EMG (Edi) on NAVA screen when the patient was in quiet phase (Non REM) of sleep, but good diaphragmatic EMG (Edi) when she was fully awake as shown in Figure 1.

Based on these findings, the infant was suspected to have congenital central hypoventilation syndrome (CCHS) which was later confirmed by the genetic study of the PHOX2B mutation.

Patient was maintained on ventilator support while sleep and she was free of any respiratory symptoms during awake. She was later discharged on long-term ventilatory home care after tracheostomy.

DISCUSSION

Central hypoventilation syndrome (also known as Ondine's curse) is a dysfunction in the metabolic control of breathing, mainly occurs during non-REM sleep.[6] The syndrome has several characteristics of central sleep apnea, in which patient's ventilation will decrease significantly or cease with no respiratory effort from diaphragm or accessory muscles. Contrary, in central sleep apnea, the resulting increase in carbon dioxide and decrease in oxygen in the blood triggers an arousal response which will transition the patient into a lesser sleep stage or awaken the patient altogether to resume breathing.[4,5]

Patients with CCHS often present with cyanotic episodes in which oxygen saturation decreases and carbon dioxide levels increase significantly with no arousal. In most cases, the physician must rule out other diagnoses with similar symptoms, such as neuromuscular and metabolic diseases. There is so far no consensus of what causes CCHS. However, a correlation recently found between patients with CCHS and a mutation of the PHOX2B gene[7] which is inherited as an autosomal dominant disorder with incomplete penetrance.[8] About 90% of all CCHS phenotypes have a mutation where there is a repeat in polyalanine sequence on the gene. The remaining 10% of CCHS phenotypes have nonsense type mutations of the gene.[9]

The apnea which is the main manifestation of this syndrome can be demonstrated clinically early by the use of the innovative NAVA technology, by detecting the absence of diaphragmatic EMG (Edi) activity during Non-REM sleep. This new technology can give the clinician an invaluable tool for instant confirmation without the need to a lengthy sleep study.

It is essential to reach an early diagnosis and provide adequate ventilatory support to prevent hypoxemic episodes. The prognosis and survival is excellent with treatment and avoidance of hypoxemia.[10]

This case report demonstrates the first reported use of NAVA technology as adjunct diagnostic tool for the diagnosis of congenital central hypoventilation syndrome in the neonatal period, utilizing diaphragmatic activity as a physiologic signal indicating an intact central neuro-respiratory drive. More studies are needed to test the feasibility and cost-effectiveness of this tool in conjunction with video EEG over the currently available sleep study methods.

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