Introduction

Congenital Central Hypoventilation Syndrome (CCHS):
- Mutation of paired-like homeobox 2B (PHOX2B) gene - key in the development of the autonomic nervous system (1,2)
- Rare: ~1000 patients known worldwide. (3)
- Patients lack autonomic reflexes that vary respiratory pattern in response to CO2 and O2
- Symptoms: small tidal volumes, periods of apnea, no respiratory rate variation, profound alveolar hypoventilation
- No response to pharmacological stimulants
- Minimal opioids
- Patients require chronic ventilatory support
- Patients lack autonomic reflexes that vary respiratory
- Patients can also have abnormalities in autonomic
- No improvement with age
- Patients require chronic ventilatory support ranging from CPAP to BIPAP to ventilator dependence
- Patients can also have abnormalities in autonomic regulation of gut motility, blood pressure, cardiac rhythm and temperature regulation(3).

The following is a case of a 9-year-old patient with CCHS, history of Hirschsprung’s Disease and esophageal dysmotility who presented for EGD and experienced significant, unrecognized hypoventilation and hypercarbic narcosis after a general anesthetic despite diaphragmatic pacing.

Case Report

Pre-operative history: SF was born full-term and made no respiratory effort at birth. She was intubated immediately and failed multiple extubation attempts as an infant due to apnea and hypoventilation. Eventually she was diagnosed with CCHS, confirmed with genetic testing. Tracheostomy was performed at 4 months and she discharged home on mechanical ventilation. Bilateral diaphragmatic pacers were placed at 6 years operatively her EGD she was dependent on her allowed her to be de...

Intra-operative care: Premedication was avoided so as not to exacerbate OSA symptoms. Propofol was given for IV induction and endotracheal intubation under direct laryngoscopy was easily performed without paralytic. The patient was maintained on a general anesthetic of sevoflurane (1.5-3.2%) and dexametomidine (1.3 mcg/kg given over 40min). Dexametomidine was chosen since it maintains respiratory function and allows for decreased opioid administration. The EGD proceeded without incident and vital signs remained stable. Deep extubation was performed due to patient’s history of severe bronchospasm and was presumed safe because of the patient’s diaphragmatic pacemakers. No narcotics or paralytics were administered. The patient was transported to the PACU with supplemental O2.

Post-Operative: In the PACU the patient maintained SPO2 of 100% with visible chest rise, obvious firing of diaphragmatic pacer and no evidence of obstruction. She was noted to be sedated for a prolonged period. After 30 minutes she continued to be difficult to arouse and CO2 narcosis was suspected. A VBG showed a PCO2 of 115 and bag-mask ventilation was started. After prolonged bag-mask ventilation the patient awoke and was then able to maintain effective ventilation without assistance. Repeat VBG showed a PCO2 of 36. The patient remained stable on home diaphragmatic pacer settings for the remainder of her stay.

Discussion

Patients with CCHS present unique challenges to anesthesiologist. They lack the intrinsic autonomic reflexes that allow humans to regulate respiratory pattern in response to oxygen and CO2 making them reliant on ventilatory support. Despite diaphragmatic pacing that was adequate in the pre-operative setting, our patient developed hypercarbia and CO2 narcosis post-operatively likely due to the sedating effects of anesthetics in the setting of baseline hypoventilation seen in CCHS.

Conclusions

Ideal peri-operative care for patient with CCHS:
- Non-sedating, short-acting anesthetics
- Minimal opioids
- Ventilatory support beyond baseline requirements post-operatively
- Vigilant post-operative monitoring of physical exam findings
- Consider end-tidal CO2 or arterial CO2 monitoring

If practitioners avoid medications that exacerbate hypoventilation and are vigilant in post-operative monitoring, anesthetic care in patients with CCHS can be optimized.

References


Sarah Moore, MD Yuan-Feng Lo, MD
Anesthesiology Critical Care Medicine, Children’s Hospital Los Angeles, Sunset Blvd Los Angeles, 90027

Functional Diaphragmatic Pacemaker Fails to Prevent CO2 Narcosis in a Patient with Congenital Central Hypoventilation Syndrome