Learning Objectives

Upon completion of this learning activity, participants should be able to:

- List common manifestations of Rett Syndrome
- Discuss the impact of these manifestations on anesthetic management
- Contrast anesthetic management choices in the OR vs austere environments such as MRI
- Formulate an anesthetic plan to care for a patient with Rett Syndrome

Introduction

Patients with Rett Syndrome present for frequent procedures requiring anesthesia care. Developmental delay, hypotonia, immature brainstem control of daytime breathing, sleep disordered breathing, autonomic instability, ECG abnormalities, seizure disorders and studies recently demonstrating increased adverse events and sensitivity to propofol contribute to creating a high risk scenario during MRI anesthesia.

Case Description

A 12 year old 40 kg girl with Rett Syndrome presented for spinal MRI. PMH was significant for neuromuscular scoliosis with 75% thoracolumbar curvature, seizure disorder, daytime periods of hyperventilation/apnea with cyanosis and a nighttime polysomnogram revealing moderate to severe OSA with AHI 13.3 and oxygen nadir of 83%. The patient was profoundly hypotonic, developmentally delayed and nonverbal. Past anesthetic history in infancy was notable for easy intubation but post-extubation apnea and desaturation requiring reintubation.

Anesthetic goals:

- Return the patient to baseline level of functioning and muscle tone quickly and completely
- Avoiding large doses of propofol
- Maintain stable and regular breathing pattern for optimal MRI images
- Use anesthetic drugs with rapid elimination times

Anesthesia Record

- Intubation: 100mg propofol + 40mg remifentanil. Easy direct laryngoscopy and placement of 6.0 ET tube
- Maintenance:
  - 50% nitrous oxide in oxygen
  - Remifentanil: 0.05mcg/kg/min
  - Propofol: 35-50 mcg/kg/min
- Ventilation: controlled ventilation for normocapnia
- Length of scan: 90 minutes
- Emergence:
  - Drips turned off at conclusion of scan
  - Time to resume spontaneous ventilation: <5 minutes
  - Time to eye-opening: 5 minutes
  - Time to extubation after return to induction room: 7 minutes
- No apnea in the immediate post-extubation period

Reports of sudden death are common.

All patients with severe OSA requiring anesthesia for MRI pose concerns for airway patency during scan as well as in the immediate post-procedural period until apnea time is regained. Patients with Rett Syndrome have additional risks including erratic breathing, autonomic instability and sensitivity to propofol.

This patient’s airway was secured due to risk of apnea and the need for regular unobstructed respiration during spinal MRI. Nitrous oxide was chosen for its low solubility and remifentanil because of its rapid metabolism by nonspecific esterases independent of infusion time. A very small amount of propofol was given to ensure amnesia during the scan. Fast elimination of anesthetic agents enabled the patient to return quickly to her preoperative level of muscle tone and cognitive function.

In conclusion, many anesthetic options are possible in patients with Rett Syndrome. An approach mindful of the physiologic disturbances tailored to the needs of the procedure and utilizing a time honored nitrous-narcotic technique worked particularly well in this patient.

Discussion

Rett Syndrome is a neurodevelopmental disease in girls caused by mutations in the MECP2 gene located on the X-chromosome encoding for CpG binding protein 2, a ubiquitous transcriptional regulator. Affected patients have profound mental disability, epilepsy and stereotypical dystonic movements. Brainstem dysfunction is nearly universal with daytime periods of hyperventilation and apnea being common. Reports of sudden death are common.

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References