Spinal Anesthesia for Cesarean Delivery in a Parturient with NKX2-1–Related (Brain–Lung–Thyroid) Syndrome

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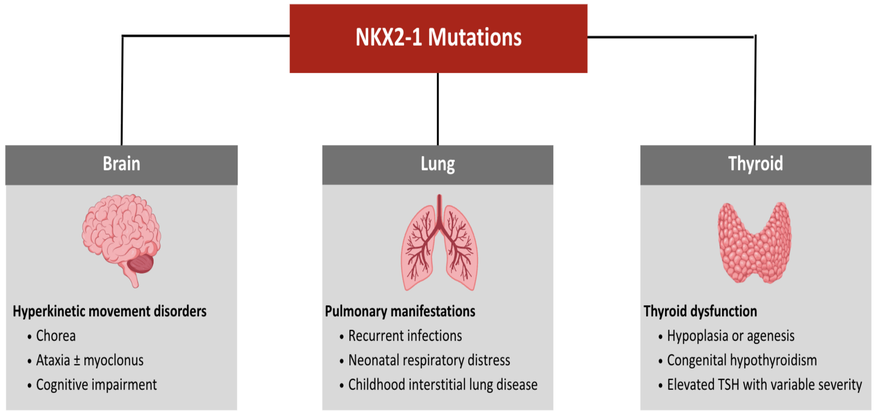
Introduction

- NKX2-1–related brain–lung–thyroid syndrome is a rare autosomal-dominant disorder (1)
- Characterized by:



- Neurologic manifestations likely arise from abnormal basal ganglia and central motor circuit development rather than from primary spinal cord or peripheral nerve pathology (1)
- Recurrent pulmonary infections are associated with surfactant dysfunction and impaired pulmonary epithelial regulation (1)
- The optimal anesthetic approach in patients with NKX2-1 syndrome remains undefined
- Neuraxial anesthesia poses a theoretical neurologic concern, whereas general anesthesia may increase pulmonary and airway risk
- We present a case highlighting anesthetic decision-making in this high-risk population

Case Presentation

Patient: G2P1 at 37.1 weeks' gestation in her 30s. Scheduled for repeat C/S. She had progressive ataxia and sensorimotor neuropathy. Recent MRI/MRA showed no acute abnormalities.
PMH: Hypothyroidism
PSH: C/S in 2013 with epidural with no complications
Meds: Baclofen, gabapentin, levothyroxine
PE: 125/95, 96, 95% SpO2, BMI 36.2 kg/m2,
Bilateral LE weakness requiring walker, sensation intact
Mallampati class III airway
L/D: WNL
Genetics: Heterozygous NKX2-1 pathogenic variant (AD)

Anesthesia plan:

- Neuraxial anesthesia was selected over GA.
- Spinal anesthesia was performed
  - 0.75% bupivacaine 12mg, fentanyl 100 mcg, and morphine 100 mcg

Anesthetic Considerations in NKX2-1 Disease

Consideration	General Anesthesia	Neuraxial Anesthesia
Pulmonary Risk	• Could increase respiratory infections	• No airway management needed
Neurologic Risk	• Neurologic risks of GA for NKX2-1 is unknown	• Might worsen neurological symptoms

Discussion

Decision-Making Framework

Spinal anesthesia was selected based on:

- Stable preoperative neurologic baseline
- Desire to minimize airway manipulation given elevated pulmonary risk profile
- Shared decision-making

Clinical Pearls

- NKX2-1 syndrome presents competing anesthetic risks across three organ systems (1)
- Pulmonary vulnerability may favor avoidance of general anesthesia (2)
- Stable baseline neurologic exam is critical prior to neuraxial placement
- Greater sensitivity to opioids and local anesthetics may be anticipated in patients with hypothyroidism, although this is based on physiology rather than syndrome-specific evidence. (3)
- Multidisciplinary planning is recommended for delivery (4)

Conclusion

- A parturient with NKX2-1–Related (Brain–Lung–Thyroid) Syndrome was successfully managed with spinal anesthesia

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