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CLINICAL VIGNETTE

Anesthetic Considerations in a Pediatric Patient with Simpson-Golabi-Behmel Syndrome

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Introduction

Simpson-Golabi-Behmel Syndrome (SGBS) is a rare, X-linked genetic disorder manifesting early in childhood with several features complicating anesthetic management. First described in 1975, the abnormalities of SGBS arise from a mutation that results in a pattern of overgrowth similar to Beckwith-Wiedemann or Sotos Syndromes.[1-3] Congenital abnormalities including macroglossia and macrosomia present familiar, but dangerous anesthetic challenges. These patients present for a wide range of surgical procedures common for conditions in this syndrome. This report describes the perioperative management of a child with SGBS requiring general anesthesia for urologic surgery.

Case Report

The patient is a 5-year-old male with SGBS scheduled for an elective diagnostic laparoscopy and likely orchiectomy for an undescended testis. The patient has intellectual delay, as well as a large tongue causing speech delay and swallowing difficulties. He receives gastrostomy tube feeding (see Figure 1). The patient exhibited distinctive syndromic facial and body features, including a large body habitus (>90% for weight), hypertelorism, and a shortened, upturned nose. He had previous surgeries as an infant for gastrostomy tube placement and orchiopexy. The patient arrived NPO on the morning of his surgery. The anesthetic plan was general anesthesia via inhalational induction of sevoflurane gas, followed by placement of a peripheral intravenous cannula and endotracheal tube for the operation.

The patient received midazolam for anxiolysis via his gastrostomy tube in the preoperative area. His mother administered the midazolam. In the operating room, mask sevoflurane was administered, and after loss of consciousness, placement of an intravenous cannula was attempted. The patient coughed during mask induction and red-tinged fluid from midazolam emerged from the mouth, indicating regurgitation of orogastric contents. The patient was placed in reverse Trendelenburg position to minimize aspiration, suctioned at the oropharynx, and given forcible positive pressure breaths via the mask with oropharyngeal airway in response to declining pulse oximeter readings. His SpO_2 decreased below 80% and he was administered intramuscular succinylcholine, a paralytic agent to prevent coughing. With the muscle relaxation and positive pressure mask ventilation, oxygen saturation returned to normal within minutes. An endotracheal tube was inserted under direct laryngoscopy, which demonstrated large, mobile lingual tonsillar tissue at the base of the tongue crowding the vallecula and glottis (see Figure 2).

The surgical case proceeded uneventfully with no cardiopulmonary instability on vital signs. He was administered ketorolac and acetaminophen for postoperative pain, and local anesthetic was infiltrated at incisional wounds prior to emergence. Following completion of the surgery, emergence from general anesthesia, and tracheal extubation, the patient was brought to the recovery room with a nasopharyngeal airway in place to prevent airway obstruction from macroglossia and residual anesthesia. The patient's mother reported flushing approximately 60 mL of water following the midazolam via the gastrostomy tube. After two hours in the recovery room, the patient exhibited no signs or symptoms of aspiration or pulmonary complications and was discharged home.

Discussion

There are two types of SGBS: type I is the classical form, whereas type II is a lethal form associated with a mutation on a different location on the X chromosome. In type I, various mutations encoding a cell surface proteoglycan glypican-3 (GPC3) result in a loss of function, presumably increasing the signaling in a Hedgehog pathway important in embryonic growth and differentiation.[4,5] GPC3 knockout mice exhibit the overgrowth that characterizes the clinical findings seen in SGBS. The X-linked recessive inheritance pattern overwhelmingly results in male patients, though rare female cases have been reported.[6-8]

The dysmorphic facial features in SGBS include a large, coarsened forehead, lips, and tongue. The tongue is widened with a midline groove. More complex airway abnormalities include cleft palate.[9] SGBS appears associated with increased risk of cardiac abnormalities, including various congenital anomalies and arrhythmias.[2,10,11] Other congenital anatomic

abnormalities increase need for surgical and anesthetic intervention. These include umbilical and diaphragmatic hernias; musculoskeletal malformations of the hand and foot, such as polydactyly and syndactyly; cryptorchidism; and duplicated renal collecting systems. Patients may have normal intelligence, though most will exhibit some disability. Patients are also at risk for embryonal neoplasms, such as hepatoblastoma, Wilms tumor, and a variety of other cancers.[1]

Our patient illustrates several clinical features of SGBS. These include a very large tongue, intellectual delay, and an undescended testis. Given lack of cooperation, anesthesia included premedication with midazolam and inhalational induction prior to placement of intravenous access. Despite inhalational induction being common in pediatric anesthesiology, lack of intravenous access delayed adequate positive pressure ventilation via mask once the patient began to cough and regurgitate. This was due to slower onset of intramuscular succinylcholine versus intravenous administration. Also, these patients are larger and heavier than similarly aged peers, with slower inhalational induction to reach deep levels of anesthesia. This allows more time for airway obstruction and complications. Furthermore, unbeknownst to the care team until after surgery, his mother flushed the midazolam through the gastrostomy tube with a small cup of water, promoting regurgitation with excess gastric volume.

Patients with macroglossia can present challenging airways for the anesthesiologist, increasing the probability of airway obstructtion and difficult endotracheal intubation. Physicians should be prepared to use oropharyngeal and nasopharyngeal airways during anesthesia administration, as well as alternative devices for airway management. These include supraglottic and laryngeal mask airways, video laryngoscopes, and bronchoscopes to assist in endotracheal intubation.

This patient's tracheal was intubated using direct laryngoscopy. Visualization was difficult due to excess, redundant tissue at the base of the tongue and lingual tonsils. At conclusion of surgery, an opioid-free pain regimen was selected to decrease likelihood of postoperative respiratory depression.

Given the abnormalities in SGBS, these patients often require anesthetic management. There are no specific, published guidelines regarding anesthesia in SGBS, though focusing on securing and maintaining a patent airway takes obvious precedence. This case illustrates how the clinical features of SGBS resulted in difficulties with oxygenation and ventilation, necessitating rapid intervention to prevent further patient deterioration.

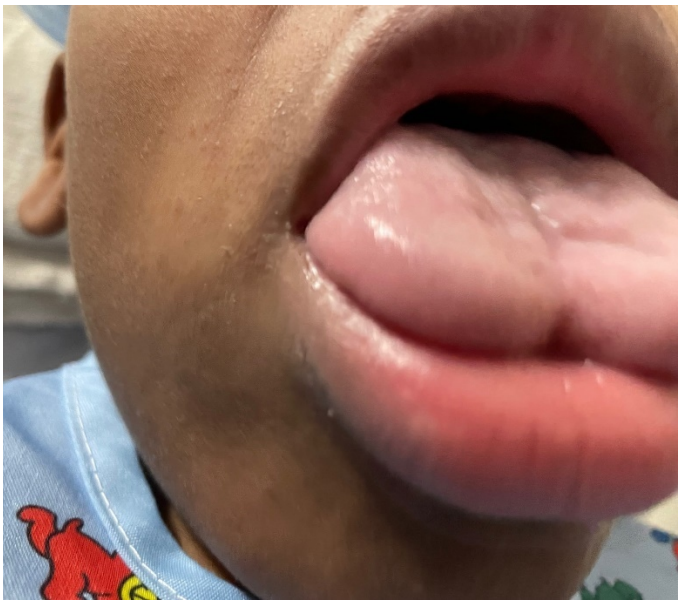

Figure 1. Patient exhibiting a large tongue with a midline groove characteristic of Simpson-Golabi-Behmel Syndrome.

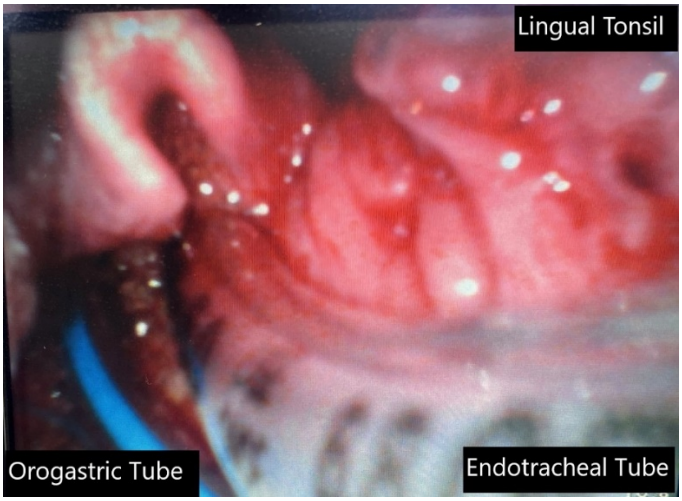

Figure 2. Laryngoscopy of the patient showing excess lingual and lingual tonsillar tissue crowding the area around the endotracheal and orogastric tube.

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