

Anesthetic Management of a Child with Simpson-Golabi-Behmel Syndrome—A Case Report

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Abstract

Simpson-Golabi-Behmel syndrome (SGBS) is an X-linked recessive condition associated with prenatal and postnatal overgrowth with visceral and skeletal anomalies. Abnormal airway anatomy may complicate the perioperative management of SGBS. However, there have been few reports regarding the anesthetic management of children with SGBS. We present the preoperative evaluation and intraoperative management of a 1.5-year-old child with SGBS undergoing plastic surgery for cleft palate. Sevoflurane-fentanyl anesthesia without a neuromuscular blocker was planned, due to the preoperative evaluation that airway management would be complicated by the macroglossia and short neck characteristic of SGBS. Fortunately, there was no difficulty in endotracheal intubation in the present patient due to careful planning of general anesthesia and meticulous airway management. Appropriate preoperative preparation for a difficult airway by the anesthesiologist can ameliorate the difficulties inherent to the anesthetic management of patients with SGBS.

Keywords

Simpson-Golabi-Behmel Syndrome, Anesthetic Management, Macroglossia

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1. Introduction

Simpson-Golabi-Behmel syndrome (SGBS), first described in 1975 by Simpson *et al.*, is an X-linked recessive condition associated with prenatal and postnatal overgrowth, macrocephaly, congenital diaphragmatic hernia, a characteristic facial appearance (large protruding jaw, broad nasal bridge, upturned nasal tip, hypertelorism, macrocephaly, macrostomia), macroglossia, supernumerary nipples, palatal abnormalities, congenital heart defects and generalized hypotonia [1] [2]. Various visceral, skeletal and neurological abnormalities are also present, including ventriculomegaly. Hypoglycemia can manifest in the newborn period. Although normal intelligence has been described, mild to severe mental retardation is common. This syndrome has variable expressivity, ranging from mild forms in carrier females to lethal infantile forms. Half of affected males die in the neonatal period. These patients are at an increased risk of tumor development in early childhood, particularly Wilms tumor, hepatoblastoma, adrenal neuroblastoma, gonadoblastoma and hepatocellular carcinoma, for which they require regular sonographic surveillance [3]. Hence, these children also frequently require operative procedures in childhood. Although abnormal airway anatomy may complicate perioperative management in this syndrome, there is only one previously published case report of the anesthetic management of SGBS in a 4.5-year-old child [4]. We present the preoperative evaluation and intraoperative management of a 1.5-year-old child with SGBS who underwent plastic surgery for cleft palate.

2. Case Report

A male neonate was delivered vaginally following a normal, full-term pregnancy after a threatened premature birth; at birth, he weighed 4148 g (+2.4 S.D.) and was 53 cm (+1.9 S.D.) tall, with Apgar scores of 9 and 10 at 1 min and 5 min, respectively. The patient had a micropenis and cleft palate. He required tubal feeding for 25 days after birth due to poor feeding secondary to the cleft plate and macroglossia. SGBS was diagnosed due to the presence of somatic overgrowth and the characteristic facial appearance, including hypertelorism, macroglossia, central tongue groove and cleft palate, as well as GPC3 gene examination. Although he had moderate overgrowth and mild developmental delay, he did not have cardiac dysfunction or abnormal glucose metabolism (Figure 1). His family history was unremarkable.

At the age of 1.5 years, plastic surgery for the cleft palate was scheduled. At this time, he weighed 12 kg (+1.8 S.D.) and was 83 cm (+1.9 S.D.) tall. Anticipating a difficult airway due to macroglossia and a short neck, sevoflurane-fentanyl anesthesia without a neuromuscular blocker was planned. General anesthesia was induced with 8% sevoflurane administered via a facemask, together with IV fentanyl (10 µg) and atropine sulfate (0.1 mg). Once an adequate depth of anesthesia was confirmed, a 4.5 mm uncuffed tracheal tube (UTT) was smoothly inserted after topical anesthesia of the airway with lidocaine, without the use of a muscle relaxant. Anesthesia was maintained with 2% - 3% sevoflurane and supplemental IV fentanyl (total 40 µg), with spontaneous respiration maintained throughout the surgical procedure. Plastic surgery for the cleft palate was performed uneventfully and a tongue stitch was taken to prevent postoperative tongue fall. Postoperatively, the endotracheal tube was removed after the patient was fully awake, with no airway obstruction after extubation. Operation and anesthesia time were 100 min and 147 min, respectively.



Figure 1. Characteristic facial appearance at the age of 2 years.

3. Discussion

Golabi, Rosen and Behmel reported several patients with SGBS syndrome, and Garganta and Bodurtha published a review of the syndrome [5]-[7]. SGBS should be differentiated from other diseases in which overgrowth occurs, such as Beckwith-Wiedemann Syndrome (BWS), Pallister-Killian Syndrome, Sotos Syndrome and Perlman syndrome [8]. SGBS is similar to BWS, which is characterized by umbilical hernia, macroglossia and a large body. Although SGBS and BWS have different causal genes, they have a similar mechanism, related to the IGF2 protein, and many clinical similarities [9]. Some patients who were previously thought to have BWS may actually have SGBS. In BWS, the rate of growth decreases slowly from childhood, such that the final height is within normal limits. However, in SGBS, overgrowth continues even after childhood, such that macroglossia can continue into adulthood [10] [11]. Patients with SGBS often attain a height of more than 195 cm.

In patients with BWS, emergency situations due to sudden respiratory obstruction and anesthetic problems such as upper airway obstruction caused by macroglossia have been reported [9] [12] [13]. In SGBS, macroglossia is the most common feature of the syndrome and difficulty in swallowing may occur at birth as well as later in the neonatal period. Therefore, airway obstruction due to macroglossia and short neck are the major problems during the anesthetic management of SGBS in early infancy, requiring appropriate preoperative planning. The principle of managing the difficult airway in this age group is to maintain spontaneous ventilation until the airway is secured [14]. Hence, a spontaneous ventilation technique that avoids the use of a muscle relaxant is recommended in these patients [14]. Due to the high risk of losing airway control, anesthetic induction with an inhalational agent such as sevoflurane, together with maintenance of spontaneous ventilation, is preferred over propofol in patients with a potentially obstructed airway [15]. If airway obstruction does occur, inhalational anesthetic administration can be discontinued and the child can be woken up. A recent study reported that use of fentanyl allowed a dose-related smooth intubation and calm emergence in children anesthetized with sevoflurane [16]. In our case, to avoid tongue-fall into the retrolingual space, which can lead to severe airway obstruction, we chose anesthetic induction with 8% sevoflurane administered via a facemask and IV fentanyl, and intubation without a muscle relaxant. We also had the Airwayscope[®] and fiberoptic bronchoscope on standby in preparation for difficult intubation. Fortunately, although bag/mask ventilation was found to be difficult, endotracheal intubation was performed without difficulty. Postoperatively, the endotracheal tube was only removed after the patient was fully awake, in order to prevent airway obstruction on waking.

4. Conclusion

We report a case of SGBS syndrome and discuss the potential anesthetic problems during surgery in patients with SGBS. The risk of upper airway obstruction due to macroglossia is one of the most important anesthetic management considerations when dealing with these patients, requiring careful preoperative airway assessment and preparations for difficult airway management before induction of anesthesia.

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