General Anesthetic Management of a Patient With Spondyloepiphyseal Dysplasia Congenita Undergoing Palatoplasty Revision

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Spondyloepiphyseal dysplasia congenita (SEDC) is a type of short-limbed dwarfism characterized by platyspondylia, delayed metaphyseal ossification, and irregularly shaped bones. Anesthetic issues in patients with SEDC have reportedly included airway stenosis caused by laryngotracheal hypoplasia, ventilation difficulty due to facial hypoplasia, and intubation difficulty attributed to microgenia. Furthermore, atlantoaxial instability can lead to cervical dislocation due to excessive or violent manipulation of the head and neck. We present the case of a 5-year-old girl with SEDC scheduled for palatoplasty revision. Airway difficulties were anticipated because of microgenia and the cervical collar she wore for atlantoaxial instability. However, mask ventilation and video laryngoscopy proved relatively easy. The patient was placed in Trendelenburg position (approximately 10°) without head tilt for surgical access. A combination of formulas based on the patient's age and height was used to determine tracheal tube size. However, the 4.5-mm oral Ring, Adair, Elwyn (RAE) tube selected resulted in 1-lung intubation when the tube bend was fixed at the lip, requiring further depth adjustment. Successful anesthetic management of this patient with SEDC incorporated several factors, including an individualized airway management plan, use of a video laryngoscope, careful posturing to avoid excessive cervical strain, and appropriate tube sizing and positioning.

Key Words: Spondyloepiphyseal dysplasia congenita; General anesthesia; Atlantoaxial subluxation; Microgenia; Case report.

Spondyloepiphyseal dysplasia congenita (SEDC) is a congenital disorder that affects growth, leading to short stature (dwarfism) and other skeletal issues characterized by platyspondylia, delayed metaphyseal ossification, and irregularly shaped bones. It is a type of osteochondrodysplasia caused by mutations in the gene for type II collagen. It may be accompanied by midface hypoplasia, cleft palate, vision issues, and hearing difficulties. In cases where atlantoaxial subluxation occurs because of hypoplasia of the odontoid process of the epistropheus (C2 vertebra), it may lead to quadriplegia or sudden death.[1] Various complications like airway stenosis due to laryngotracheal hypoplasia and intubation difficulties due to microgenia should be considered during the anesthetic management of patients with SEDC. For those patients with atlantoaxial instability, violent or excessive laryngeal manipulation during intubation and excessive movement of the head/neck during surgery may cause cervical vertebral dislocation. In addition, endotracheal tubes for patients with short stature should be cautiously selected because of the risk of overinsertion and 1-lung intubation.

In the present study, we report a case of a child with SEDC who underwent palatoplasty revision under general

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anesthesia. Written consent was obtained from the patient's parents for the publication of this report.

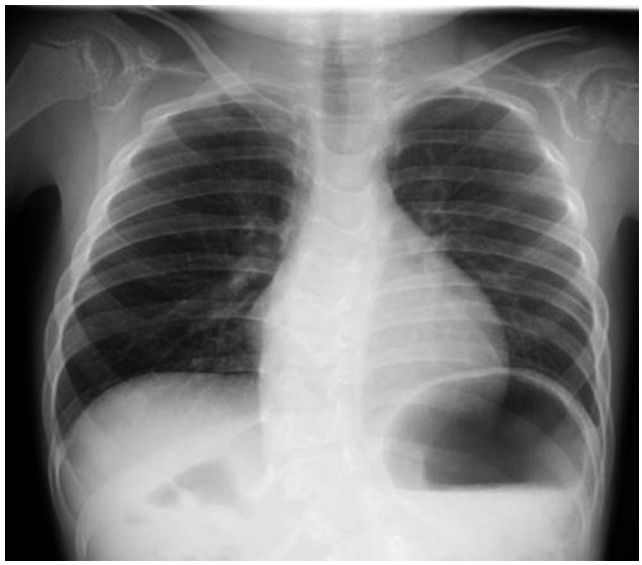
CASE PRESENTATION

A young girl 5 years and 8 months of age (height 87 cm, weight 13 kg, body mass index 17.2 kg/m^2) with SEDC was scheduled to undergo palatoplasty revision under general anesthesia. Although the possibility of shortened lower limbs was identified during fetal ultrasonography, no abnormalities were detected by congenital metabolic screening. The patient was delivered without complications, although she was noted as having a cleft palate. However, she was examined 2 months after birth at a local clinic with the chief complaint of dwarfism and was diagnosed with SEDC. She underwent palatoplasty under general anesthesia at our hospital at 1 year and 11 months of age but developed a palatal fistula afterwards.

Upon presentation for the palatoplasty revision, her family denied any other pertinent medical comorbidities, noteworthy family medical history, current medications, or allergies. She had been diagnosed with atlantoaxial instability, for which she was wearing a cervical collar daily and could not bend her head backwards. Physical evaluation revealed facial and physical findings indicating microgenia and short stature, respectively. Preoperative posterior-anterior (PA) chest radiograph revealed no apparent morphologic aberrations in her respiratory tract (Figure 1), whereas a lateral cervical radiograph revealed hypoplasia in the odontoid process of the epistropheus. Although the patient had a Mallampati class I airway, we anticipated difficulties securing her airway because of microgenia and the inability to extend her neck during intubation. Her other routine preoperative testing, including standard preoperative blood tests and a 12-lead electrocardiogram, indicated no abnormal findings.

While developing the anesthetic plan for this patient, an awake intubation was deemed impracticable considering her age. Therefore, the following was planned: anesthesia would be induced slowly using sevoflurane. If mask ventilation proved insufficient once the anesthetic depth intensified past the excitation stage, the patient would be woken up emergently and the operation terminated to avoid the risk of deficient ventilation and intubation. If mask ventilation proved feasible, a muscle relaxant (rocuronium) would be administered and the patient intubated using a McGRATH video laryngoscope (McGRATH Mac, Covidien) fitted with a size 2 blade. If intubation could not be done using the McGRATH Mac, intubation using a flexible fiberoptic scope would be attempted. If intubation seemed not feasible, the patient would be woken up and the operation terminated. This plan was explained fully to the patient's parents beforehand.

Figure 1. Preoperative PA chest radiograph.

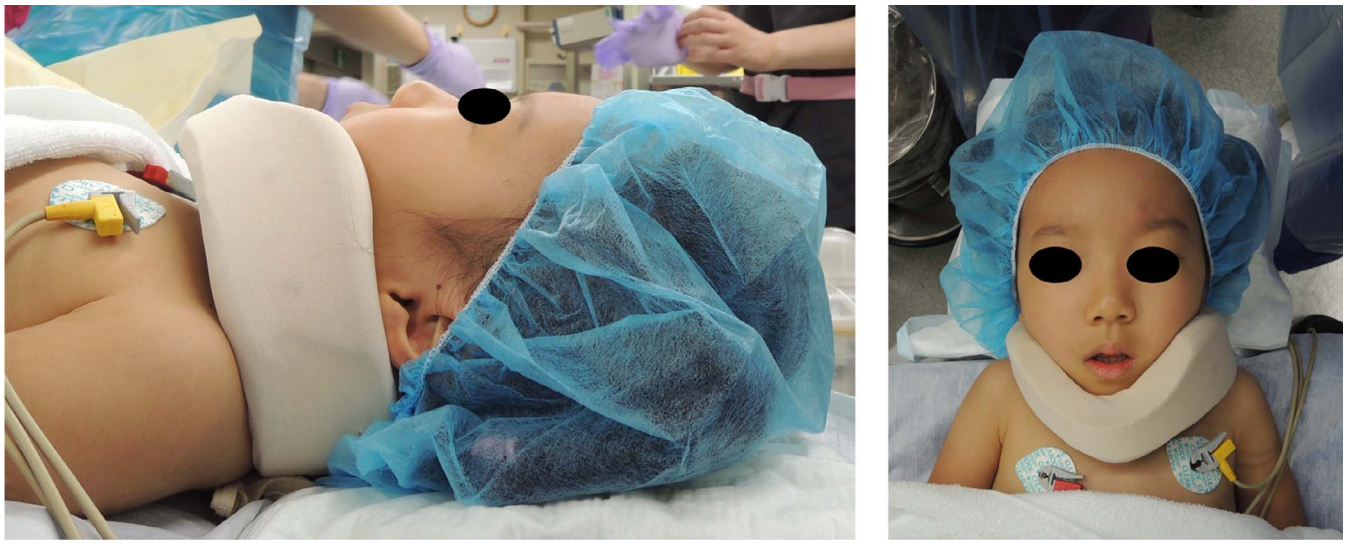


No apparent morphologic aberrations were noted involving the respiratory tract.

To protect the patient's unstable cervical spine, she was brought into the operating room for induction while wearing her daily cervical collar (Figure 2). After sufficient preoxygenation 6 L/min, the patient was induced using sevoflurane 8%. Once the depth of anesthesia was past the excitation stage, mask ventilation was possible using a jaw thrust. After securing a 24-gauge intravenous (IV) cannula in her left forearm, an IV bolus of rocuronium 10 mg was administered and continuous infusion of remifentanil 0.2 μg/kg/min was initiated. Notably, the cervical collar remained in place to help ensure the patient's head was not extended. After muscle relaxation was achieved, oral intubation was attempted using the McGRATH Mac with a disposable size 2 blade along with a stylet inserted into a pediatric-sized 4.5-mm, cuffed, oral RAE (Microcuff, Avanos Medical) endotracheal tube (ETT). The Cormack-Lehane classification using the McGRATH Mac was grade 2, indicating that visualization of the airway and intubation was easier than expected.

Following intubation, 0.5 mL of air was inflated into the ETT cuff. Because fixation of the ETT at a preformed bend (length 15 cm) resulted in 1-lung ventilation, the insertion depth was set at 14 cm to ensure 2-lung ventilation. With a sponge inserted between the bend in the ETT and the patient's lip to preclude 1-lung ventilation, the ETT was taped in place along the middle of the lower lip (Figure 3) after which the cervical collar was removed. General anesthesia was maintained using oxygen 1 L/min, air 2 L/min, sevoflurane 2%, and remifentanil 0.1-0.2 μg/kg/min. Lidocaine 1% with 1:200,000 epinephrine was administered via

Figure 2. Lateral and frontal views of the patient prior to intubation.

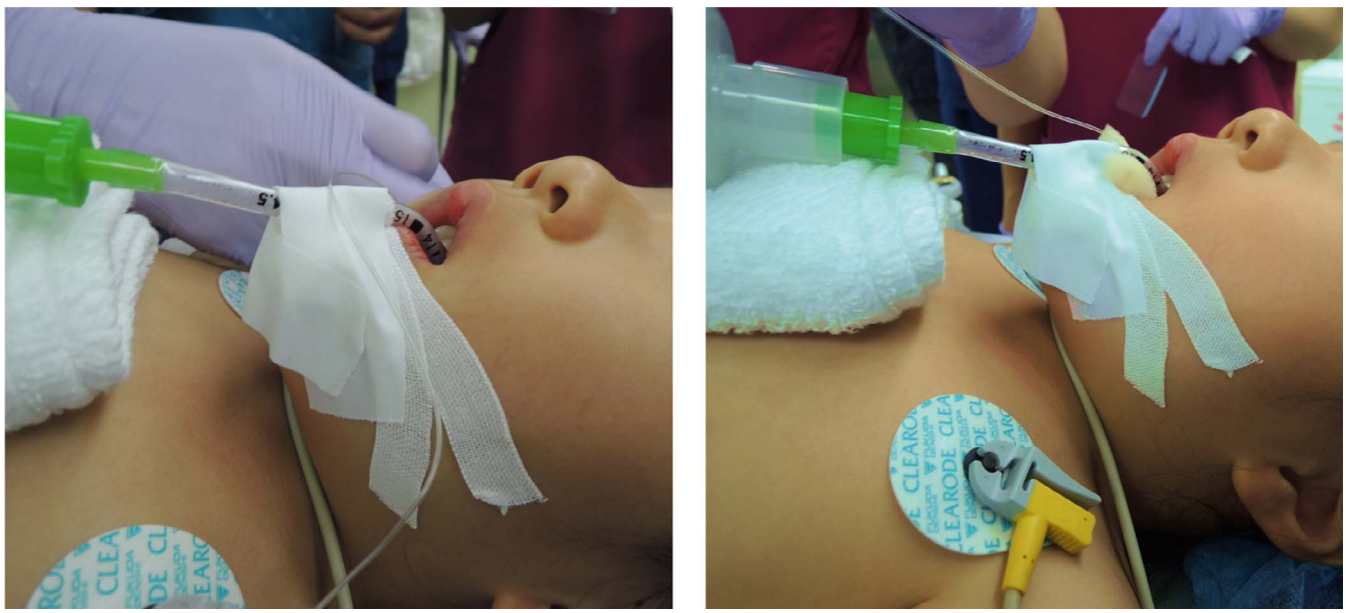


Her cervical spine was protected by the cervical collar.

palatal infiltration for local anesthesia; total dose was lidocaine 30 mg, epinephrine 15 µg.

Intraoperatively the patient was placed in Trendelenburg position at approximately 10° while avoiding extension of her head. Intraoperative vitals were stable, and an IV bolus of fentanyl 20 µg was administered 10 minutes before completing the operation to prevent agitation upon awakening. Additionally, an acetaminophen suppository 200 mg was administered for postoperative analgesia. The patient's cervical collar was reattached postoperatively before she emerged from anesthesia. Because her train-of-four (TOF) ratio upon completion of the operation was 0.4, an IV bolus of sugammadex 30 mg was given to reverse the neuromuscular paralysis. After the TOF ratio stabilized at approximately 1.0, the patient swiftly awoke without any intense body movements. She was awake and responsive prior to extubation, and the ETT was removed after confirming sufficient ventilation and oxygenation. Her vital

Figure 3. Lateral views of the endotracheal tube (ett) positioning postintubation.



Left: Before the sponge was inserted. Right: After the sponge was inserted between the ETT bend and the patient's lip to preclude 1-lung ventilation.

signs remained stable postoperatively. The operation lasted 1 hour and 2 minutes, the anesthetic time was 2 hours and 12 minutes, and the amount of bleeding was 10 g.

DISCUSSION

Spranger and Wiedemann[1] reported SEDC for the first time in 1966 as a rare genetic disorder primarily characterized by postnatal dwarfism and hypoplasia in the proximal limb bones. Patients with SEDC have short stature and can develop kyphosis and scoliosis, which results in thoracic deformities and genu valgum (a knock-knee deformity). Other physical findings likely to impact anesthetic management are midface hypoplasia, laryngotracheal hypoplasia, and microgenia. Patients with SEDC typically start experiencing gait disorders of varying severity with age because of the destruction of epiphyses. This appears to be due to underdevelopment and fragmentation of the epiphyseal bone and cartilage.[2] It appears that most cases of SEDC are de novo/novel mutations that are then inherited in an autosomal-dominant fashion.[3]

In contrast to SEDC, achondroplasia is the most common cause of short-limbed and disproportionate dwarfism and is characterized by macrocephaly and long-bone shortening of proximal upper and lower extremities.[4] Achondroplasia has an estimated incidence rate[5] of 1 in 20 000 to 30 000 live births whereas SEDC has an estimated incidence[1] of 1 in 100,000, indicating that SEDC is an extremely rare growth disorder.

As mentioned above, various factors must be considered while managing an anesthetic for patients with SEDC. First, the possibility of airway stenosis and difficulties with mask ventilation and intubation due to laryngeal hypoplasia should be considered. Some case reports describe successfully performed endotracheal intubation using an Airway Scope (AWS; Pentax) in patients with microgenia when laryngeal manipulation was infeasible.[3,6] Although the patient's preoperative chest radiograph in the present case did not reveal any apparent findings of airway stenosis, securing the airway was expected to be challenging owing to microgenia and strict limitations in being able to extend her neck. Although an oropharyngeal airway was prepared to help counteract the anticipated difficulties during mask ventilation, sufficient mask ventilation was achieved in this case with only a jaw thrust and chin lift. In selecting a video laryngoscope for tracheal intubation, we initially considered using an AWS, as it can facilitate tracheal intubation without any need of head extension. However, we selected the McGRATH MAC because it also could feasibly facilitate visualization of the airway and intubation without head extension, and we were most accustomed to its operability and usability. Consequently, we achieved tracheal intubation without encountering any problems.

The second issue was that violent tracheal intubation procedures might cause cervical dislocation in the presence of the patient's unstable atlantoaxial joint. In fact, one case of fatality due to cervical injury of a patient has been reported.[7] Therefore, it was essential to preoperatively confirm the safety of extending her neck by verifying her routine range of motion the day before surgery and consider using a cervical collar for fixation. Securing a patient's head and neck in a neutral position and then placing them into the Trendelenburg position has been previously reported as a successful way to perform surgery requiring that type of positioning while avoiding the risks of excessive extension.[8] In the present case, anesthesia was induced while the patient was wearing her daily cervical collar, and it was not removed because of surgical access until the airway was secured to protect her unstable cervical spine.

During a palatoplasty, sufficient head extension is required for visual and surgical access of the operative field. However, because the patient's head could not be extended intraoperatively because of her unstable atlantoaxial joint, she was placed into Trendelenburg position intraoperatively, which enabled sufficient access for the surgical team. The bed angle was restricted to approximately 10° to avoid the risk of increasing intracranial and airway pressures because of diaphragmatic elevation while in the Trendelenburg position.

The third issue was selection of an ETT appropriate for patients with short stature. According to one report, many cases involving short patients used an ETT with an actual size at least 1.0 mm smaller than the predicted size considering the patient's age.[9] At our hospital, a cuffed ETT is selected for all patients because it rarely requires a size change to create an appropriate seal, unlike uncuffed ETTs, and has a low incidence rate of aspiration.[9] It is believed that a combination of multiple predictive formulas should be employed to determine the type of ETT for a child patient rather than a single formula. In the present case, a combination of formulas factoring in the patient's age was employed to select an appropriately sized cuffed ETT. We used the Motoyama formula (inner diameter [ID] [mm] = 3.5 + age [years]/4) and the Knine formula (ID [mm] = 3 + age [years]/4), which yielded values of 4.75 and 4.25 mm, respectively. Consequently, as a mean of these values, a 4.5-mm Microcuff ETT was selected.

Owing to the need to fix the ETT on the median position of the lower lip during palatoplasty, we utilized a pediatric-sized, preformed, oral RAE ETT because it was impossible to secure a standard oral ETT in such a position without negatively impacting access for the surgical team. However, as previously reported,[4] morphologic aberrations are often noted in SEDC; therefore, using a smaller-diameter ETT than the size calculated based on the patient's age

would have been suitable. As the Microcuff ETT resulted in 1-lung intubation when affixed at the preformed bend in the tube, the length of the ETT selected for this case was too long and required further adjustment such as a sponge inserted between the ETT bend and the patient's lip. Thus, in this case we realized the importance of choosing an ETT that was smaller than the calculated size while selecting a pediatric oral RAE ETT. Nevertheless, one must balance various factors, including avoiding an overly wide or narrow (in diameter) ETT as well as a tube that is too long or short, when selecting an appropriately sized ETT that fits the individual patient's physique.

CONCLUSION

We report the successful anesthetic management of a female patient with SEDC who underwent palatoplasty revision. Because of anticipated intubation difficulties and existing cervical instability, we took various measures such as developing an individualized anesthetic/intubation plan, using a McGrath video laryngoscope, and placing the patient in Trendelenburg position after securing the airway. Although the ETT we selected resulted in a 1-lung ventilation, we were able to adapt it successfully by using a sponge to adjust its depth. Consequently, we were able to successfully manage the general anesthesia for this patient with a medical history complicated by SEDC.

DISCLOSURE

None of the authors of this article have any conflicts of interest to disclose.

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