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Case Report

Anesthetic management of a pediatric patient with Rett syndrome - A case report -

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Rett syndrome is a neurodevelopmental disease that almost always affects female patients. It is caused by mutations in MeCP2 in the majority of cases. Patients diagnosed with Rett syndrome may experience serious adverse events even with smaller amounts of medication for sedation and anesthesia. The major anesthetic concerns associated with Rett syndrome are lack of cooperation, abnormal continuous limb movements, abnormal respiratory control, difficult positioning secondary to scoliosis, and altered sensitivity to painful stimuli. Because of the risks caused by these problems, anesthesiologists should be aware of the specific anesthetic concerns of patients with Rett syndrome in order to safely administer anesthesia. Here, we describe the management of a pediatric patient diagnosed with Rett syndrome.

Key Words: MeCP2 mutations, Postnatal neurological disorder, Rett syndrome.

Rett syndrome is a postnatal neurological disorder that almost always affects female patients. The estimated cumulative incidence of Rett syndrome in Australia is 10 per 100,000 females by the age of 12 years, and it is considered to be the second most common cause, after Down syndrome, of severe mental retardation in females [1]. Clinical features include episodic hyperventilation, breath holding, air swallowing, bruxism, night laughing, screaming spells, early growth arrest of feet, vasomotor hyper-reactivity of feet, neurogenic scoliosis, altered sensitivity to painful stimuli, and intense eye communication [2].

Safe management of general anesthesia in these patients requires consideration to the development of seizures, a disturbed breathing pattern with hyperventilation and periodic apnea, and difficult intubation due to scoliosis [3]. We describe here the anesthetic management of a pediatric patient with some of these features who was diagnosed with Rett syndrome.

CASE REPORT

A 39-month-old girl with Rett syndrome was scheduled to undergo elective surgery for left distal femur fracture (greenstick fracture). She had normal pre- and perinatal periods, and her development was normal during the first 6 months. However, she could neither sit well unsupported nor crawl until 12 months of age. The patient's body weight and height were in the normal range, but her head circumference was below the 3rd percentile in comparison with others of her age group. Progressive mental retardation and several seizure events were observed after the age of 12 months, and seizure medication was initiated. At the age of 24 months, the patient visited Seoul National University Hospital, where she was diagnosed with Rett syndrome. Following her diagnosis, she underwent standing physical therapy twice a week, and a left femur fracture occurred accidentally during the course of that therapy.

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The patient was admitted to the rehabilitation department of our hospital. In collaboration between the orthopedic and rehabilitation specialists, it was decided that she would undergo conservative treatment for the femur fracture.

The patient's body weight was 9.8 kg (below the 5th percentile) and height was 82 cm (below the 5th percentile). During physical examination, she showed tenderness in the left femur. Her Mallampati score was class I and her mouth was wide enough to allow the insertion of two fingers. She was floppy in general and often showed wringing movement of the hands and feet. Her seizures were well controlled with valproate. Her blood pressure was maintained within the range of systolic 90–95 mmHg/diastolic 60–65 mmHg and heart rate was maintained within 120–125 beats/min. There were no abnormal findings in preoperative laboratory data, electrocardiogram (ECG), or chest radiography. No anatomical or functional abnormalities had been observed on previous echocardiography, which was performed in the process of diagnosis of Rett syndrome.

We planned to perform assisted ventilation with mask bagging considering the short operation time and potential delayed recovery of a patient with Rett syndrome. Upon arrival in the operating room, she was calm and cooperative. Noninvasive blood pressure monitoring, 3-lead ECG, and a pulse oximetry monitor were placed. The patient's blood pressure and heart rate were 98/62 mmHg and 100 beats/min. Anesthesia was induced with thiopental sodium 50 mg, after which mask ventilation was begun with O_2 6 L/min and 2 vol% of sevoflurane. No muscle relaxant was injected because assisted facemask ventilation was sufficient to maintain $ETCO_2$ within 35 ± 5 mmHg. Assisted facemask ventilation was performed with O_2 1.5 L/min, air 1.5 L/min, and sevoflurane 1.5–2.0 vol% when the patient was unable to perform spontaneous breathing. Her blood pressure was maintained within the range of systolic 80–85 mmHg/diastolic 55–60 mmHg, and heart rate was maintained within 120–125 beats/min.

The operating time was 10 minutes and there were no complications. Endotracheal intubation was not required because manual reduction was performed successfully without open manipulation. After the surgery, the patient's breathing was assisted with O_2 100%. About ten minutes later, spontaneous respiration was fully recovered to allow maintenance of SpO_2 of 100% without assisted ventilation. The patient was transferred to the postanesthesia recovery room with no adverse events. Ten minutes after arrival in the recovery room, the patient was able to maintain eye contact with her mother. Her blood pressure was maintained within the range of systolic 90–95 mmHg/diastolic 60–65 mmHg and heart rate was maintained within 120–125 beats/min, and she was transferred to the ward after 20 minutes of monitoring in the recovery room. The patient remained in the ward for 4 days without any additional complications, and was then discharged.

DISCUSSION

This case deals with a pediatric patient who was diagnosed with typical Rett syndrome. Rett syndrome is a neurodevelopmental disease that is often caused by mutations in MeCP2; these mutations can be found in 96% of individuals with typical Rett syndrome [4]. Diagnosis is considered when postnatal deceleration of head growth is observed. Clinical features associated with typical Rett syndrome include regression of purposeful hand skills and spoken language, with aggravation of gait abnormalities and hand stereotypies [5].

According to the recently revised clinical criteria for the diagnosis of Rett syndrome, the main criteria are: 1) Partial or complete loss of acquired purposeful hand skills; 2) Partial or complete loss of acquired spoken language; 3) Gait abnormalities: impaired (dyspraxic) or absence of ability; and 4) Stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing and washing/rubbing automatisms. The exclusion criteria are: 1) Brain injury secondary to trauma (peri- or postnatally), neurometabolic disease, or severe infection that caused neurological problems; and 2) Grossly abnormal psychomotor development in the first 6 months of life [4,5]. Our patient met all four main criteria of the diagnostic criteria and did not meet the two exclusion criteria.

Clinical staging is useful in understanding Rett syndrome. This system, which was established in 1986, classifies the disease onset into 4 stages. Stage I (early-onset stagnation) begins by 6 to 18 months of age with loss of acquired developmental skills, particularly involving speech and hand movements. Stage II (developmental regression), which can last for many months, begins by 1 to 4 years of age, with further loss of acquired skills and mental deficiency. Stage III (pseu-

dostationary period) can last for many years and is marked by a plateau of developmental regression, with some improvement in communication skills. Stage IV (late motor deterioration) is characterized by more severe neuromuscular disability, with deterioration in ambulation, and commonly includes progressive scoliosis [3].

As detailed above, it is necessary to use caution with sedation and anesthesia in individuals diagnosed with Rett syndrome, because they show severe aggravation of disease with age. Close observation is needed because of the tendency of these individuals to experience episodic hyperventilation and breath holding even with smaller amounts of medication than normally required. Preparation for perianesthesia vomiting and potential aspiration is needed because of possible gaseous distention resulting from bloating and air swallowing. Moreover, many experts believe that immature brainstem function could lead to unexpected sudden death and prolonged apnea during sedation and anesthesia. In addition, preparation for difficult intubation and restrictive lung disease owing to significant scoliosis is needed from stage IV [3].

A previous case regarding the anesthetic management of a patient with Rett syndrome was reported in our country [6]. A 19-year-old female, who was in stage IV of Rett syndrome, underwent ophthalmic surgery under general anesthesia. The patient had scoliosis and had been bedridden for 5 years. On examination, the thyromental distance was 3.5 cm and the airway was normal with the exception of trismus and excessive secretions. Because the trismus was expected to improve after the injection of rocuronium, direct laryngoscopy was attempted. However, while the patient was fully relaxed, her mouth opening was very limited. Therefore, fiberoptic-guided orotracheal intubation was performed. Following surgery, weaning from the ventilator was attempted. However, because the patient's recovery was slow and complicated by episodes of shaking, she was transferred to the intensive care unit, and it took a day for her to regain full consciousness.

The patient in this case was in stage II of Rett syndrome and was scheduled to undergo closed reduction for left femur fracture. It was possible to perform controlled ventilation with inhalant agents alone because the patient showed no self respiration during anesthetic maintenance. Low tidal volume ventilation was applied to prevent gaseous distention of the stomach. In addition, her oral cavity was monitored several times, and secretions were carefully suctioned. Because the surgery was performed without muscle relaxation, recovery of spontaneous respiration was fairly rapid. After observing recovery of spontaneous respiration, we inserted a Levin tube into the patient's oral cavity to drain residual gas naturally. The patient was transferred to the postanesthesia care unit after a few minutes of observation.

In conclusion, the major anesthetic concerns associated with Rett syndrome are lack of cooperation, muscle wasting, abnormal continuous limb movement, abnormal respiratory control, difficult positioning secondary to scoliosis and chest deformity, vasomotor instability, metabolic abnormalities (increased lactic acid levels), and altered sensitivity to painful stimuli [2]. Rett syndrome is a severe progressive deterioration of the central nervous system in the young child. Since the diagnosis of Rett syndrome is becoming more common [3], anesthesiologists should be aware of these anesthetic concerns associated with Rett syndrome in order to safely administer anesthesia. In addition, anesthesiologists should inform caregivers about possible aggravation of the disease with aging, as well as adverse events that may be caused by sedation and anesthesia.

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