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CASE REPORT

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Rett Syndrome and anaesthetic management: a case report

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Abstract
Rett syndrome is a progressive neurological disorder that occurs only in females and it manifests with mental retardation, seizures, movement disorders, autistic behavior, and abnormal breathing. A 4.5-year-old female child with Rett syndome underwent dental restoration under sedoanalgesia. Her physical examination was normal except for sterotyped movements. We recommend that anesthesiologists should have knowledge about this disorder's characteristics, complications, potential associated problems, and important aspects of anesthetic drug selection.

Keywords: Anaesthesia, dental surgery, rett syndrome

Introduction

Rett Syndrome (RS) is a rare neuroprotective disorder involving only females, which affects noradrenaline (NA) innervation in cerebral cortex and hippocampus by altering the functions of cells in locus coeruleus by mutation of a gene encoding methyl-cytosine-guanosine (CpG) binding protein 2 (MECP2) located on chromosome X [1] Its prevalence is 1/10.000. In addition to the loss of cognitive, motor, and social abilities, it is characterized by epilepsy, stereotypic hand movements, autistic behavior, abnormal respiratory patterns, gastroesophageal reflux, nutritional issues, and severe scoliosis. Although female children with RS demonstrate a normal or near normal development until the 6th to 18th month of life, they lose their skills thereafter and then enter a long-term non-development phase [2].

In this case report we aimed to stress some important points to be remembered by anesthesiologists in persons with Rett syndrome.

Case Report

A 4.5-year-old, 25 kg female child with Rett syndrome was scheduled to undergo outpatient dental restoration under sedoanalgesia. She underwent preoperative evaluation. She was mentally retarded; and although her consciousness was open, she lacked cooperation and orientation. She had mental and motor developmental retardation and was not able to walk. Her past history was notable for stereotypic movements that had begun at the age of 6 months and increased in intensity by 12 months of life, which were later attributed to Rett syndrome. She had no other illnesses. Airway examination was unremarkable except fort he caries in teeth. Auscultation of the heart and lung was normal with murmur and additional sound in cardiac examination, with no ral, rhanchi in respiratory examination, respectively. Her head and neck examinations were all normal. She can provide adequate mouth opening; she did no have any seizure activity; and her electrocardiography (ECG) was normal. Her hemoglobin level was 10.4 g/dl and serum electrolytes were within normal limits. In the operating room, her baseline vitals were as the following: heart rate 120/min, noninvasive blood pressure 85/55 mmHg, and peripheral oxygen saturation (SpO2) 98%. No premedication was administered and, in operating room, following the administration of 0.1 mg/kg demizolam and 1 mg/kg propofol iv the operation was started. She was kept on spontaneous ventilation with nasal oxygenation. Then, propofol 1 mg/kg was added. Intraoperative period was uneventful, with no period of desaturation. After the completion of the 20-min long operation, the patient was taken to the recovery room and monitored until after she re-gained full motor motion abilities. She was discharged after two hours of follow-up.

Discussion

As the rarely encountered Rett syndrome causes life-threatening complications, the anesthetic care of patients with this syndrome is clinically important.

RS is characterized clinically by autistic behavior, mental retardation, respiratory problems, loss of speech and manuel skills, holding breath, oral-motor dysfunction, scoliosis, autonomous dysfunction and somatic developmental impairment. The incidence of sudden death from RS has been reported as high as 22-

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25% . Cardiac end-organ involvement and abnormalities of the central respiratory control have been defined as the causes of sudden death. The potential cardiac etiological factor for sudden death is the prolonged QT interval. Patients with Rett syndrome have an increased prevalence of prolonged QT interval and T wave abnormalities compared to the age-matched population. Cardiac involvement worsens as disease stage increases. Therefore, preoperative evaluation should include 12-lead ECG. Additionally, as prolonged QT interval is also associated with increased sympathetic tone, bisoectral index (BIS) monitorization is prudent to use during both the induction stage and throughout the operation. Prior studies have shown that volatile anesthetic agents isoflurane, desflurane, and sevoflurane, as well as ondansetron with antiemetic properties, prolong QT interval whereas propofol and remifentanyl do not affect it. Moreover, antiarithmic properties of magnesium can be utilized when arrhythmia occurs [3].

The other cause of sudden death in RS is an abnormal respiratory pattern ranging from hyperventilation to apnea caused by the loss of cerebral nervous system (CNS) control. In addition, neck and head examination during preoperative evaluation is also important since intubation procedure may be complicated by problematic intubation caused by difficult airway control associated with limited mouth opening, micrognatia, and limited neck movements. Additionally, respiratory dysfunction, chronic aspiration, and recurrent pneumonia secondary to hypotonia may necessitate re-intubation and mechanical ventilation at the postoperative period. Therefore, rapid sequence intubation may be considered with the use of an appropriate mask, rapid-onset neuromuscular blocker, and cricoid compression. Although rocuronium is a good option, it may produce a prolonged response in patients with hypotonic CNS disorders. Succinylcholine may cause hyperkalemia in patients with neurodegenerative disorders [3-4]. Jerome et al encountered severe masseter spasm with the use of succinylcholine for rapid sequence intubation in a patient with Rett syndrome [5]. Furthermore, succinylcholine creates a tendency for prolonged QTc interval in patients with fatal arrhythmias [6]. Administering sugammadex with incapsulation property with rocuronium may provide rapid neuromuscular recovery. Kawasaki et al achieved an uneventful recovery by establishing a BIS value of 30 to 50 and using sugammadex in a patient with Rett syndrome who was scheduled for laparoscopic fundoplication and gastrostomy operation and in whom difficult airway was anticipated due to trismus and micrognatia [7]. Endotracheal intubation can be achieved with propofol-remifentanyl combination, without using muscle relaxant [8].

Patients with RS are excessively sensitive to sedatives, opioids, and volatile anesthetic agents. Tofil et al reported that patients with RS had a lower anesthetic dose requirement compared to the control group and suffered prolonged apnea at the postoperative period [9]. Regional anesthesia should be preferred for suitable operations in patients with Rett syndrome since it reduces opioid, anesthetic requirement and postoperative respiratory depression [10].

Seizure is a common co morbid condition in patients with RS. Therefore, therapeutic anticonvulsive level should be determined and optimized preoperatively in order to control postoperative seizures. Barbiturates, propofol, and volatile anesthetic agents have potent anticonvulsive properties [11]. Since hypokapnia causes seizure-like activity, caution should be exercised during anesthesia induction and high-frequency ventilation [12].

Since up to 50% of affected patients may have orthopedic deformities including scoliosis and/or multiple joint contractures and even malpositioned vascular formations, arterial cannulation and venous access may become difficult [3].

Our patient had non-advanced Rett syndrome characterized by the absence of seizure activity, micrognatia, limited mouth opening, orthopedic deformity. She also lacked pathological cardiac and respiratory patterns that cause the greatest number of deaths. Despite having a non-advanced stage, our patient was still well managed both peri- and postoperatively with the help of a detailed and careful preoperative examination, avoiding opioids, carefully observing in terms of apnea and using propofol and midazolam because of not cause prolonged QT interval and implement outpatient anesthesia. We had a shortcoming. We could have utilized BIS monitored which was allowed us to follow anesthetic depth and to titrate the effect of anesthetic agent.

With the present case report we aimed to help anesthesiologists successfully manage all stages of operations like a maestro in patients with Rett syndrome and other neurodegenerativedisorders.

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