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Case Report

Anesthesic Approach in Patients with Williams Syndrome

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Abstract

Williams-Beuren (WBS) Syndrome, commonly known as Williams Syndrome, is a congenital and multisystemic disease described in 1961-1962. It has cardiac pathologies that may lead to sudden deaths in anesthetic approach and craniofacial anomalies that may cause difficult airway. In this case, we aimed to present our anesthesia approach in a patient who was scheduled for operation by Gynecology and Obstetrics Department because of labia minor hypertrophy.

Keywords: Anesthesia; General Anesthesia; Williams Syndrome

Introduction

Williams-Beuren (WBS) Syndrome, commonly known as Williams Syndrome, is an autosomal dominant, congenital, multisystemic disease defined in years 1961-1962 with a prevalence of 20,000-50,000 live births [1-3]. It was showed by Ewart et al. in 1993 that Williams syndrome is caused by microdeletion of elastin gene on chromosome 7q11.23 [4,5]. Congenital heart diseases such as supravalvular aortic stenosis and peripheral pulmonary arterial stenosis accompanied by characteristic facial appearance and dental anomalies are considered to be a multisystemic syndrome due to its characterization developmental delay, genitourinary and endocrinological symptoms, mental retardation, infantile idiopathic hypercalcemia [6,7]. Low birth weight and growth retardation are the main symptoms and typical facial features include microcephaly, broad forehead, flattened nose root, short palpebral wrinkles, wide mouth with marked lower lip, micrognathia, operiorbital fullness and epicantial cantlants [8].

Case Report

The patient who was scheduled for surgery by the Gynecology and Obstetrics Department due to Labium minus hypertrophy being a 12-year-old and weighing 31 kg girl and she was diagnosed with Williams syndrome. Preoperative evaluation included peri-facial appearance, wide mouth, dental disorders and micrognathia. In the ECO done on the patient, it was determined that the patient had adequate EF and mild aortic regurgitation and aortic stenosis. Blood pressure was calculated as 105/55 mmhg, heart rate was calculated 130 beats / min and body temperature was calculated as 36.8 ^{0}C Her vital signs were stable and no pathological findings were found in the blood laboratory values studied. A more non-invasive approach was planned due to the short duration of the planned operation as well as possible difficult intubation and ventilation. The patient was taken to the operating room and the vascular access was opened with 22G intraket. Fluid replacement was started to be isotonic 80cc / min. ECG, non-invasive blood pressure, peripheral oxygen saturation, end-tidal CO_2 monitoring were performed. 1 mg midazolam was administered for sedation. In addition to face masks of various sizes, airway and LMA, the Clarus video system with different numbers of blades and endotracheal tubes was prepared for a possible difficult airway. After three minutes of preoxygenation, anesthesia induction with 1.5 mcg / kg fentanyl and 3 mg / kg propofol was applied to the LMA patient number 3. There was no problem during and after this procedure. The patient's chest expansion was adequate and $EtCO_2$ square waveform was observed in normal pattern. Tidal volume and respiration rate were adjusted to 32-35 mmHg for $EtCO_2$. Mixture of 50-50% O_2-NO_2 and infusion of propofol 75 mcg / kg / min was used on the maintenance of anesthesia. There was no deterioration in vital signs during the 20-minute procedure. At the end of the case, the patient was woken up without any complications. The patient was sent to the service after the following of PACU. Pain control was provided with paracetamol in the postoperative period. The patient was discharged from the hospital on the third day

without any problems.

Discussion

In this case report, we aimed to present our anesthesia experience on a patient with Williams syndrome who was scheduled for operation due to labium minus hypertrophy. As it is known, there are many system involvements on patients with Williams syndrome and these patients should be evaluated preoperatively, an appropriate intraoperative anesthesia method should be determined and a good analgesia control should be provided postoperatively.

The incidence of congenital heart disease in these patients is 60-80% and 93% of them are diagnosed at the age of one [9-10]. For this reason, firstly, cardiac research should be done. The main cause of Supravalvular Aortic Stenosis (SVAS) and Pulmonary Artery Stenosis (PAS) on these patients is the microdeletion of the elastin gene, the major artery wall protein [9]. Cardiac pathologies including SVAS and PAS are the leading cause of sudden death in this patient group [11]. Myocardial ischemia, low cardiac output and ventricular arrhythmias are the main causes of these sudden deaths and often result in unsuccessful resuscitation. Also case series including 19 cases by Bird et al. support this situation [12]. In addition, Gupta P et al.'s failed resuscitation in cardiac arrest during contrast-enhanced CT scan on a child with Williams syndrome supports this situation [13]. Considering all of these, a comprehensive evaluation including ECO in addition to a good cardiac examination in the preoperative period should be applied to the patients with Williams syndrome. For this purpose, we performed a cardiac examination on our patient and planned to perform an ECHO for further evaluation of the patient. For further evaluation, we consulted the Pediatrics Department and we operated with endocarditis prophylaxis as an outcome of the consultation.

In this patient group, there are micrognathia known as typical face appearance, large mouth and dental disorders. This may complicate mask ventilation and intubation of patients during the induction [14]. In a case by Medley et al., it was reported that a 7-month-old girl could not be intubated with cuffless intubation tube suitable for her age and weight and then developed laryngeal edema [6]. Therefore, it is obligatory to have difficult airway instruments available in cases with general anesthesia. In the presented case, there was no difficulty during airway control and LMA application.

One of the other affected systems is the musculoskeletal system. In these cases, muscle weakness, joint contracture, muscle fat storage and increased muscle fiber sizes showed an increase. Therefore, unexpected responses under the influence of muscle relaxants may be observed as a result of any disturbance at the neuromuscular junction [15]. Also, cases of masseter muscle spasm due to neuromuscular agents have been reported [16]. If a muscle relaxant is planned to be used on these patients, the muscle relaxant should be titrated and accompanied by neuromuscular monitoring (TOF). In the presented case, no muscle relaxant was applied to the patient.

Hypercalcemia, abdominal pain, vomiting, constipation may be observed as a result of ELN gene deletions on patients with Williams syndrome. At the same time, seventh Hypercalcemia may occur in parallel with the effect of the L-type voltage-dependent calcium channels on the chromosome. This may be associated with the formation of malignant hyperthermia [17]. Patel and Harisson used halothane and sucamethonium on a patient with Williams syndrome and reported masseter muscle spasm [16]. However, Jannu V administered 0.8-1 MAC sevoflurane with oxygen-air mixture for inguinal hernia surgery to an 8-year-old boy and no complications were observed [18]. Therefore, if general anesthesia is preferred in these cases, good planning should be made and total intravenous anesthesia (TIVA) should be preferred especially to avoid malignant hyperthermia. In this case, we provided general anesthesia infusion and maintenance with propofol. We did not encounter any complications.

As a result, multisystemic involvement is present in patients with Williams syndrome. These patients should be investigated well in the preoperative period. In order to prevent sudden deaths, a wide range of cardiological examinations should also be performed including ECO. In order to avoid difficult airway that may occur parallel to the typical facial appearance, the necessary instrumentations should be prepared or regional anesthesia should be used. In addition, a preoperative preparation after doing necessary preparations is required on this patient group which is prone to malignant hyperthermia.

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