Ambulatory Anesthesia Management for a Pediatric Patient with Kabuki Make-Up Syndrome: Case Report

Kabuki Make-Up Sendrom'lu Bir Pediatrik Hastada Ambulatuar Anestezi Yönetimi

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ABSTRACT Kabuki Make-up Syndrome is a rare, sporadic congenital syndrome. The main features are classic facial appearance and dermatologlypic pattern with mental retardation, joint laxity, short stature and prominent finger pads. The facial features include long palpebral fissures, epicanthal folds, depressed nasal tips and tooth abnormalities. Patients can present renal and skeletal abnormalities. Congenital heart defects such as a single ventricle, double outlet right ventricle or transposition of the great vessels are common in Kabuki Syndrome. Difficult airway, cardiovascular problems, malignant hyperthermia, hypotonia, genitourinary abnormalities and epilepsia may increase the risk associated with general anesthesia. Every day more cases are described attracting the attention to Kabuki syndrome. In this case report, we aimed to discuss the anesthesic management of a 28-month-old patient with Kabuki Syndrome.

Key Words: Kabuki syndrome; monitoring, ambulatory; coronary angiography

ÖZET Kabuki Make-up Sendromu ender rastlanan sporadik, konjenital bir hastalıktır. Mental retardasyon, kısa boy ve eklem gevşekliğinin eşlik ettiği klasik yüz görünümü ve dermatoglipik patern bu sendromun ana özelliklerini oluşturmaktadır. Yüz karakteristikleri uzun palpebral fissürler, epikantal katlantılar, deprese nazal uç ve diş anomalilerini içermektedir. Hastalar renal ya da iskelet anomalileriyle karşımıza çıkabilirler. Tek ventrikül, çift çıkışlı sağ ventrikül ya da büyük damar transpozisyonu gibi kardiyak defektler Kabuki Sendromu'nda sıktır. Hastalığa eşlik eden zor havayolu, kardiyovasküler problemler, malign hipertermi, hipotoni, genitoüriner hastalıklar ve epilepsi genel anesteziyle ilişkili risk oranını artırabilir. Kabuki Sendromu'na dikkat çeken vakaların sayısı gün geçtikçe artmaktadır. Bu olgu sunumunda Kabuki Sendrom'lu 28 aylık hastanın anestezi yönetimini tartışmayı amaçladık.

Anahtar Kelimeler: Kabuki sendromu; izlem, ambulatuar; koroner anjiyografi

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Kabuki Make-up Syndrome is a sporadic congenital syndrome. The estimated frequency of this syndrome is about 1/32 000. The main features are classic facial appearance and dermatologlypic pattern with mental retardation, joint laxity, short stature and prominent finger pads.[1] The facial features include long palpebral fissures, epicanthal folds, depressed nasal tips and tooth abnormalities.[2] Patients can present cardiovascular, renal and skeletal abnormalities. Cardiac defects contribute approximately one-third of all patients.[3] Anesthetic management of Kabuki Make-up Syndrome has been reported previously in the literature. However

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the use of laryngeal mask airway is rare. To our knowledge, this case is the first ambulatory anesthesia report for pediatric patients with Kabuki Make-up Syndrome.

CASE REPORT

The patient consent form was obtained from parents. The patient was 28-month-old, 8.8 kg, American Society of Anesthesiologists (ASA) II, female, diagnosed with Kabuki Make-up Syndrome and scheduled for diagnostic coronary angiography under general anesthesia.

One year ago she underwent correction surgery for total anomalous pulmonary venous return and congenital diaphragmatic hernia operation. Fiberoptic orotracheal intubation was performed before this surgical correction. The patient had pectus excavatum, however she had no pulmonary, renal or liver diseases. In her physical examination, Mallampati score was class 4 with limited mouth opening. She had distinctive facial features including epicanthal folds, prominent ears, high-arched palate and tooth abnormalities (Figure 1). She had no heart murmur and lung sounds were clear bilaterally. Chest radiograph showed cardiomegaly and hyperinflation of left side of lung (Figure 2).

She was monitorized including standard DII derivation electrocardiography, noninvasive blood pressure, heart rate and pulse oximetry. General anesthesia was induced with intravenous (iv) 2 $mg^{-1}kg^{-1}$ propofol, iv 0.6 $mg^{-1}kg^{-1}$ rocuronium bromide and alfentanil 10 $mcg^{-1}kg^{-1}$. Anesthesia was maintained with 2% sevoflurane and nitrous oxide in oxygen (33%). Size 1.5-laryngeal mask airway has been performed easily. There has been no important changes in monitorized values perioperatively and angiography has been completed one and half hour later. The muscle paralysis was reversed with iv 0.03 $mg^{-1}kg^{-1}$ neostigmine with 0.015 $mg^{-1}kg^{-1}$ atropine at the end of the surgical intervention. Following the removal of laryngeal mask airway the patient was transferred to the post anesthesia care unit. Postoperative course was also uncomplicated and the patient was discharged from hospital 6 hours after the procedure.

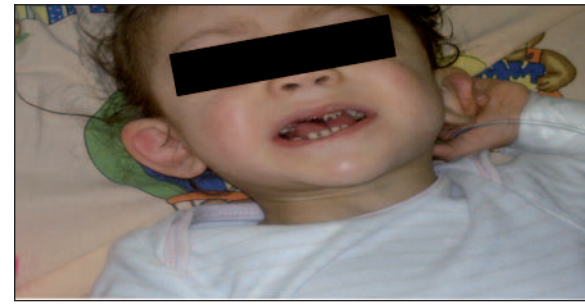

FIGURE 1: Class 4 Mallampati score with limited mouth opening.

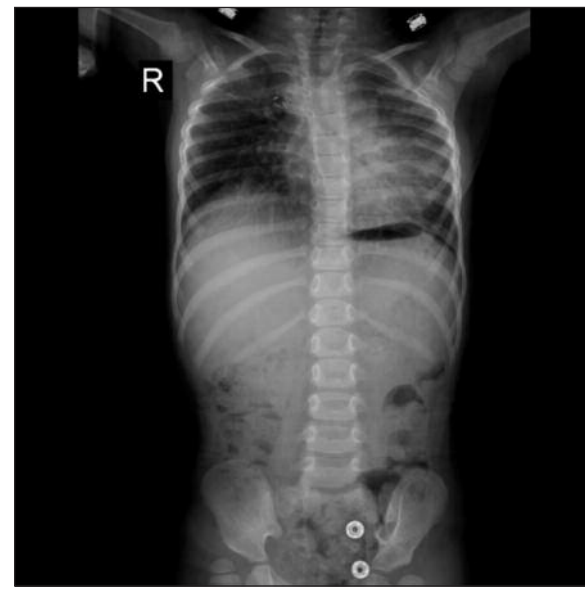

FIGURE 2: Chest radiograph with mild cardiomegaly and hyperinflation of the left lung.

DISCUSSION

Kabuki Make-up Syndrome was first described simultaneously in Japanese patients in 1981.[4] The etiology of Kabuki Make-up Syndrome is not well defined but most of the cases are sporadic and probably with an autosomal dominant mutation with variable expressivity but some patients have shown chromosomal abnormalities.[5] Although Kabuki Make-up Syndrome may occur in other ethnic groups including Northern European, Brazilian, Filipino, Vietnamese, East Indian or Chinese population, most reported cases are still Japanese chil-

dren.[6] Currently there is no confirmatory genetic test and also there is no consensus regarding diagnostic criteria. All of the patients have typical facial features and they can help diagnose Kabuki Make-up Syndrome. In this case, the patient was characterized by depressed nasal tip, arched eyebrows, and conspicuous ears. Furthermore several diseases accompany Kabuki Make-up Syndrome. Secundum atrial septal defect, membranous ventricular septal defect and cleft palate are samples. Congenital heart defects including ventricular septal defect, atrial septal defect, patent ductus arteriosus or congenital anomalies of the aortic arch were reported to be seen in 31% of 62 children with Kabuki Syndrome.[7]

Muscular hypotonia is common for these patients. Despite the normal muscle biopsies, hypotonicity of muscles and prolongation of neuromuscular-blockade may increase the risk of malignant hyperthermia.[8] Total intravenous anesthesia was reported as an alternative to inhalation anesthesia and the complications were expected to be less. Remifentanil has been used safely instead of inhalation anesthesia without complication.[4] Sivaci et al. preferred sevoflurane, midazolam and opioids for anesthesia, because propofol may cause an epileptic episode.[9] We used both alfentanil and inhalation anesthetics. By this way hemodynamic instability did not occur.

Anesthesia for rare syndromes may generate anxiety to the anesthetist responsible for administering anesthesia because of unique challenges during surgical procedures.[10] The risk of anesthesia is related with cardiovascular, neurological, urogenital and skeletal muscle abnormalities. Therefore the patients need careful preoperative evaluation that brings along successful perioperative management.

Airway problems that affect anesthetic management are micrognathia and cleft palate. Therewith, anesthesiologist needs to be especially prepared for difficult intubation. Casado et al. did not use any neuromuscular blocking agents because of child's hypotonia and possible difficult airway.[6] Preparation for fiberoptic tracheal intubation, tracheostomy or retrograde intubation is also suggested. Before anesthesia, we thought to encounter possible difficult airway management. The characteristic facial appearance, high Mallampati score and limitation of mouth opening of the patient caused this. However we maintained the airway with laringeal mask airway more easily than we expected. During the recovery from anesthesia, the absence of laryngospasm, bronchospasm or respiratory depression enabled the early discharge of this patient.

Because of hypotonia children with Kabuki Make-up Syndrome are liable to become obese in adolescent period. This may cause to obstructive sleep apnea which is presented with postoperative respiratory complications. Scoliosis which is seen in 35% of patients may affect respiratory function. Our patient did not have either obstructive sleep apnea or scoliosis. Yucel et al. also used laryngeal mask airway without any problem as we used.[8] They have mainly given remifentanil with sevoflurane during the induction and maintenance of their patient.

In conclusion, every day more cases are described attracting the attention to Kabuki Syndrome. Due to their abnormalities, patients have a greater need to surgical procedures thereby they are in the need of anesthesia. We are of the opinion that careful preoperative assessment should be considered by anesthesiologists in order to overcome perioperative complications.

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