

Anesthetic Management of a Patient with Ellis-van Creveld Syndrome

Ellis-Van Creveld Sendromunda Anestezi Yönetimi
Anesteziyoloji ve Reanimasyon

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Özet

Ellis-van Creveld Sendromu (EVC) otozomal resesif geçişli kompleks bir genetik sendromdur. Disproporsiyone boy kısalığı, postaksial polidaktili, ektodermal displazi ve konjenital kalp hastalığı tetradı ile karakterize olsa da pulmoner, renal, hepatik organ etkilenmesi de görülebilmektedir. EVC tanılı hastaların kardiyak ya da el ve dişlerdeki anomalileri düzeltmek için anesteziye gereksinimleri olabilir. Biz bu olgu sunumunda diş çekimi ve dolgu planlanan EVC tanılı hastamızdaki anestezi yaklaşımımızı sunmayı amaçladık.

Anahtar kelimeler: *Ellis-van Creveld Sendromu, genel anestezi, zor entübasyon*

Abstract

disproportionate short dwarfism, postaxial polydactyly, ectodermal dysplasia and congenital heart disease, pulmonary, renal and hepatic organs may also be affected. Patients diagnosed with EVC may require anesthesia for the restoration of cardiac, hand or teeth abnormalities. In this case report, we aimed to present our anesthetic approach in a patient with EVC who was scheduled for tooth extraction and filling.

Keywords: *Ellis-van Creveld syndrome, general anesthesia, difficult intubation*

Introduction

Ellis-van Creveld (EVC) syndrome, also known as chondroectodermal or meso-ectodermal dysplasia, is a complex genetic syndrome with autosomal recessive inheritance¹. Classic syndrome is characterized by the tetrad of disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia and congenital heart disease which mostly affects teeth and nails. Anesthetic care is frequently required during operations of cardiac pathologies, and also hand and teeth abnormalities in these patients. Patients with EVC have the risk of difficult intubation and ventilation due to their oral and dental anomalies. In addition, other organ pathologies especially cardiac pathologies are important in terms of perioperative morbidity and require a careful anesthetic management²⁻⁴.

In this report, we aimed to discuss our anesthesia experience in a patient diagnosed with EVC.

Case Report

A 6-year old patient diagnosed with EVC syndrome presented to our clinic for anesthesia preparation for dental surgery. According to the history received from the family, the patient was the first and only child of the family and was born by normal vaginal delivery. The patient had been operated at the age of 9 months due to atrioventricular septal defect (AVSD) and received enapril 2.5 mg (1x1). The patient had no any active complaint and known allergy.

On physical examination, her weight was 11.5 kg, height was 89 cm, and both were under 5 percentiles according to age. There was bilateral symmetric extremity shortness, 6 fingers in both hands, 6 toes in the right foot and also

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syndactyly. Intraoral examination performed by dentists revealed developmental insufficiency in the maxilla and mandible, irregular maxillary alveolar cleft and attached frenulum. Furthermore, along conical incisors and microdontic molars, there were numerous number and shape anomalies such as taurodontism and hypodontia. Mouth opening and neck movements were normal.

Respiratory sounds and preoperative chest radiograph were normal and there was no complaint related to the pulmonary system. Cardiac auscultation revealed rhythmic heartbeats and 1/6 systolic murmur in meso-cardiac focus. Grade 1 left and right AV valves failure and 60% ejection fraction was observed on the preoperative echocardiogram of the patient which was evaluated by the department of cardiology. The patient was evaluated as having a hemodynamically stable heart by the cardiologists.

Preoperative hemogram, renal and hepatic tests and coagulation profile were normal. Our patient was assessed according to the American Society of Anesthesiologists as ASA 2 risk patient and prepared for the operation accordingly. The family was informed about the anesthesia method that would be performed and the possible risks. The family gave written consent.

Modified Yale Preoperative Anxiety Scale (mYPAS) score of the patient was 10 before premedication⁵. The patient was given 0.5 mg/kg oral midazolam for premedication. After 30 minutes, mYPAS become 6 and the patient underwent routine monitoring (electrocardiogram, pulse oximetry, non-invasive blood pressure). Baseline vital findings were; blood pressure: 87/46 (65), Nb: 102, SpO₂:100. Following preoxygenation with 100% oxygen for 3 minutes, anesthesia induction was initiated. Inhalation induction using 100% oxygen and 8% sevoflurane (4MAC) was administered. After intravenous access was established from dorsal of the left foot with a 26 gauge cannula, 2 mg/kg propofol was infused. The patient underwent nasal intubation with no 4 spiral tube without using neuromuscular blocking agent using video laryngoscopy (STORZ DCI video laryngoscope, Karl Storz, Tuttlingen, Germany, blade no:2). The maintenance was provided by 50% oxygen-air mixture, 2.5% sevoflurane and remifentanyl (1 µg/kg/dk). Hemodynamic parameters of the patient were stable during the operation. IV 0.5 µg/kg fentanyl was administered for postoperative analgesia. The maintenance was terminated at the end of the operation, which lasted 90 minutes, and the patient was extubated without any problem. The patient was followed up in post-anesthesia care unit (PACU) until Modified Aldrete Score reached 10 and discharged with recovery on the postoperative 1st day⁶.

Case Discussion

Ellis-van Creveld syndrome is a rare autosomal recessive congenital disease. It is mostly seen in Amish population, who lives in a certain region of the USA, and its incidence is approximately one case per 5000 live births. Its incidence for the general population is 1/60000-100000 live births⁷. EVC is seen equally in both women and men⁸. The disease is caused by mutations in EVC1 and EVC2 that are localized in the short arm of 4p16 chromosome^{9,10}. No genetic test was performed in our patient.

Besides, characteristic primary defects related to EVC, other defects associated with the other organ systems also affect perioperative anesthetic care in these patients¹¹. Congenital heart diseases are seen about 50-60% of patients and are the major factor shortening lifetime¹². Endocardial fusion defects like ostium primum atrial septal defect (ASD), ventricular septal defect (VSD) and mitral valve anomalies are commonly seen cardiac defects¹³. Therefore, preoperative evaluation should involve echocardiography or cardiac catheterization. Patients with congenital heart disease who will undergo non-cardiac surgery should receive antibiotherapy in order to prevent subacute bacterial endocarditis. Our patient received treatment of 50 mg/kg oral amoxicillin before the procedure.

Difficulties with airway management in these patients increase perioperative morbidity. Oral and dental malformations like cleft palate/lip make airway management challenging¹⁴. Maxilla, mandibula and frenulum

malformations can make mask ventilation difficult¹¹. For these reasons, preoperative airway evaluation of the patient must be performed. All preparations for difficult airway should be made in the operating room. Although, mouth opening and neck movements were natural in our patient, we think that difficult ventilation and intubation could be seen due to insufficient development of the mandibula and malformed teeth in our patient. Therefore, different sizes of masks, airways, laryngeal masks, intubation tubes and video laryngoscopy were kept available in the operating room. Induction was initiated in our patient following preoxygenation and ventilation difficulty did not occur. The patient was intubated with video laryngoscope without any problem.

Patients with EVC may have different thoracic deformities that can lead to respiratory problems and difficulties experienced in mechanical ventilation may cause barotrauma. Taking airway under control rapidly prevents the risk of aspiration in these patients. Preoperative anamnesis should be taken whether these patients have chronic lung disease or infection and should be consulted if necessary¹³. In preoperative medical history and physical examination of our patient, there was no respiratory findings.

Preoperative renal functions should be investigated since renal anomalies including nephrotic syndrome, nephrolithiasis, and renal medullary dysplasia can be seen^{15,16}. Hepatic failure progressing to hepatic transplantation may be seen, and especially hepatic function should be investigated in patients with liver dysfunction findings^{15,17}. Preoperative values of our patient were normal and the results were within normal range in the postoperative follow-up. Although, there is no exact information about the selection of anesthetic agents, no contraindication has been described about using inhaler agents or neuromuscular blocking agents in these patients. Abeles et al. safely used midazolam, ketamine, fentanyl, dexmedetomidine, nondepolarizing neuromuscular blocker (cisatracurium) and inhaler agents (sevoflurane and desflurane) for intubation in a patient scheduled for cardiovascular surgery¹¹. Gupta et al. safely used midazolam, fentanyl, acetaminophen, nondepolarizing neuromuscular blocker (atracurium) and inhaler agent (sevoflurane) for inserting laryngeal mask airway in a patient scheduled for surgery due to polydactyly¹³. We did not use neuromuscular blocker since we did not need muscle relaxation during intubation and tooth extraction and filling. We used midazolam in premedication, propofol and sevoflurane in induction, remifentanyl and sevoflurane in maintenance and fentanyl in postoperative analgesia.

In conclusion; patients with EVC should be well evaluated preoperatively. Especially, patients with cardiac disease should be evaluated with echocardiography by cardiologists, prophylaxis for infective endocarditis should be planned, chronic pulmonary disease should be questioned, and hepatic and renal functions should be assessed. Necessary precautions should be taken for difficult ventilation and intubation, and a video laryngoscope should be kept available in the operating room. We believe that midazolam, propofol, sevoflurane, remifentanyl and fentanyl provide safe anesthesia in these patients.

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