Case Report

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Anesthesia Management in Robinow Syndrome (A Case Report)

Hacer Yeter[1], Murat Bagci[1], Aydin Firincioglu[1], Sitki Nadir Sinikoglu[1], Kerem Erkalp[1], Aysin Selcan[1]

[1]Istanbul Bagcilar Training and Research Hospital, Department of Anesthesiology and Reanimation, Istanbul-Turkey

Corresponding author:
Hacer Yeter,
Istanbul Bagcilar Training and Research Hospital, Department of Anesthesiology and Reanimation, Istanbul, Turkey
Phone: +90-212-440-4000
E-mail address: dr.hacer@yahoo.com.tr

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ABSTRACT

Robinow Syndrome (RS) is a rare disease characterized by anomalies in the face, head, external reproductive organs, and spine segmentation. The three main symptoms of the syndrome are fetal face appearance, genital hypoplasia, and gingival hyperplasia. Fifteen percent of the cases have congenital heart defects. Short neck, large tongue, and airway problems due to a structural disorder of the face may be observed. In this paper, we present our anesthesia practice in a case that had been diagnosed with RS.
Keywords: anesthesia, Robinow Syndrome

ÖZET

Robinow Sendromu'nda anestezi yönetimi (olgu sunumu)

Robinow Sendromu (RS), yüz, baş, dış üreme organları ve omurga segmentasyonlarında anormalliklerle karakterize, nadir görülen bir hastalıktır. Sendromun üç ana belirtisi, fetal yüz görünümü, genital hipoplazi ve gingival hiperplazidir. Olguların %15'inde doğumsal kalp kusurları var. Yüzün yapısal bozukluğuna bağlı kısa boyun, büyük dil ve havayolu problemleri görülebilir. Bu yazıda, RS'lu bir olgudaki anestezi pratiği sunuldu.
Anahtar kelimeler: anestezi, Robinow Sendromu

Introduction

Robinow syndrome (RS) occurs in two forms, autosomal dominant RS (DRS) and autosomal recessive RS (RSS). It is a rare disease characterized by anomalies in the face, head, external reproductive organs, and spine segmentation.

Main symptoms of RS include brachymelia, hemivertebrae, dysmorphic face ("fetal face"), genital hypoplasia, micropenis, clinodactyly, camptodactyly, hypoplastic fingers, and moderately short body structure. Cardiac defects (especially right ventricle outlet obstructions) and hepatosplenomegaly can be observed (1).

It may be necessary to administer anesthesia to patients being followed with RS for reasons such as skeletal or cardiac anomalies. Short body structure, large tongue, and potential airway problems may cause difficulties in intubation according to the structural disorder of the face.

Aim of this report is to present our anesthesia administration in a case with RS.

Case report

A stoma enlargement operation was planned for a male case (7 months old, 5640 gr) who had been hospitalized due to respiratory distress and subsequently received tracheostomy. It was learnt that the case was being followed for RS and had been treated for choanal atresia at the age of 3 months and undergone tracheostomy when he was 4 months old. In preoperative examination of the patient, facial appearance peculiar to RS, Mallampati IV, tachypnea, common wheezing were present. An orogastric probe for feeding was present in the patient. Laboratory values, electrocardiography (ECG) and echocardiography (ECHO) were normal. In posterior-anterior lung radiography, thoracic scoliosis and vascular shadowing had a tendency of increase. The patient was considered to be ASA II. After 8 hours of fasting, the patient was taken to the operating room; monitoring was made by ECG, noninvasive blood pressure, peripheral oxygen saturation (SpO_2), and body temperature. Heart rate was 188 beats/min, blood pressure

was 150/60 mmHg, SpO_2 was 86%. The patient was preoxygenated for 3 minutes with 100% oxygen via tracheostomy cannula. After the induction of anesthesia with 1-8% sevoflurane, a 24G peripheral venous catheter was placed into a vein on the dorsal side of the right hand and intravenous fluid administration was begun. After 1.5 mcg/kg fentanyl injection intravenously, the tracheostomy cannula no. 3 was removed and a reinforced cuffed endotracheal tube (ETT) of 3.0 mm internal diameter (ID) inserted. For anesthesia maintenance, 2% sevoflurane in a 50:50 O_2/air mixture was administered. A neuromuscular agent was not used. The patient had no bleeding after the operation and his postoperative heart rate, blood pressure values and SpO_2 values were normal. For tracheostomy, a cuffless cannula no. 4 was inserted after an operation of 20 minutes. The placement of the cannula was verified by auscultation and capnograph. The lungs were sufficiently aspirated from within the tracheostomy cannula. Sevoflurane was stopped and the patient was awakened (Figure 1). The patient was kept in the recovery room for nearly two hours and then sent to the ward. In the postoperative period, no complications were observed and the patient was discharged without any problems after 24 hours.

Written Informed Consent was taken from the patient's relatives.

Discussion

Genetically, RS can be autosomal dominant, autosomal recessive, or heterogeneous. The most important manifestations are mutations in the autosomal recessive gene ROR2. ROR is part of the tyrosine kinase family located at the cell surface, and it plays a role in the formation of the skeleton, the cardiovascular and the genital system. The ROR_2 gene is located on chromosome9q22. Its frequency is 1:500.000, and it is equally observed in both sexes (2). In these patients, surgical operations may be needed for vertebral, orofacial, and dental anomalies (2). Since there are facial and dental form disorders as well as a short neck and short body structure, there may be problems in ventilation and airway control with a mask. Due to dysmorphic facial appearance, necessary preparations need to be made for a possible difficult intubation (3,4). Congenital airway anomalies such as extrathoracic airway obstruction and functional airway obstruction in these cases may require a tracheostomy (5).

In our case, due to the choanal atresia at the age of 3 months, a surgical operation was undertaken and tracheostomy was performed when he was 4 months old. Atrial septal defect, aortic coarctation, tetralogy of Fallot, severe pulmonary stenosis or atresia and tricuspid atresia are congenital heart defects being reported with a prevalence of 15% in these patients. Due to pulmonary and cardiac complications, nearly 5-10% of the cases die in early childhood (2). In our case, tachycardia was present but cardiac auscultation, ECG and ECO were normal.

In patients with RS, in the presence of renal cyst and obstructive uropathies in addition to genital hypoplasia,

Figure 1: Postoperative view of the patient (Robinow Syndrome).

analysis of preoperative basal blood urea nitrogen, creatinine and electrolyte need to be conducted, and if any pathological result is found in these tests, additional examinations including ultrasonography have to be conducted (2). In our case, laboratory values were normal.

Chest wall movements and respiratory functions of patients with RS may be affected due to pectus excavatum, scoliosis, and costal anomalies. In serious cases, coughing and secretion-cleaning capabilities can be disrupted, which increases the tendency of respiratory system infections. In our case, tachypnea and common wheezing were present. In posterior-anterior lung radiography, thoracic scoliosis and vascular shadowing had a tendency of increase. Besides, current vertebral anomalies in these patients may make the administration of regional anesthesia difficult (6). We performed the tracheal stoma revision operation under general anesthesia. Eating difficulties and esophageal reflux are reported (7). In our case, an orogastric probe for feeding was present in the patient. Laboratory values, ECG and ECHO were normal. In posterior-anterior lung radiography, thoracic scoliosis and vascular shadowing had a tendency of increase.

Mental and motor development of the cases is generally normal. Mental retardation and development retardation are only reported in 20% of the patients. Although macrocephaly is a frequently observed finding, it does not create a risk factor for the development of retardation. Although structural anomalies related to the central nervous are known to occur system, only one case with cortical dysplasia has been reported (2). In our case, mental and motor development were normal.

In conclusion, in RS, especially in the presence of half-face anomaly and micrognasia, the anesthetist should be careful about airway difficulties. Preoperative evaluation of the airway, respiratory, renal and cardiac function should be checked. Since intraoperative complications are not only related to possibly difficult airways but include cardiac anomalies, preparations are to be made for these issues.

Contribution Categories	Name of Author
Follow up of the case	H.Y., M.B., A.F.
Literature review	H.Y., M.B., S.N.S., A.S., K.E., A.F.
Manuscript writing	H.Y., M.B., S.N.S. A.S., K.E.
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