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CASE REPORT

Anesthesia Management in An Adult Patient with Desbuquois Syndrome

Murat Tümer[1]
ORCID: 0000-0001-9132-9992

Hazar Kökten[1]
ORCID: 0000-0001-7146-816X

Aysun Ankay Yılbaş[1]
ORCID: 0000-0001-6196-3191

Fatma Sarıcaoğlu[1]
ORCID: 0000-0002-2706-5651

[1]Department of Anesthesiology and Reanimation, Faculty of Medicine, Hacettepe University, Ankara, Turkey.

Corresponding Author: Murat Tümer
Department of Anesthesiology and Reanimation, Faculty of Medicine, Hacettepe University, Ankara, Turkey.

E-mail: dr.m.tumer@gmail.com

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ABSTRACT

Desbuquois syndrome (DS) is a rare disease that can be accompanied by a difficult airway. In the literature, there are usually case reports about the mortal forms in childhood. We wanted to share our anesthesia experience in an adult patient with DS who underwent orthopedic surgery. We used videolaryngoscopy for intubation with coronavirus precautions. Adult patients with DS can be managed safely with adequate difficult airway preparation.

Keywords: Desbuquois syndrome, difficult airway, anesthesia management

INTRODUCTION

Desbuquois Syndrome (DS) is an autosomal recessive syndrome characterized by short stature, joint dislocation, and defects in the hand and foot bones [1,2]. There are two types of DS, Type-1 (mild form) with a mutation in the CANT-1 gene and Type-2 (severe form) with XYLT1 mutation. Airway management can be challenging due to short neck and facial deformities. In our knowledge, there is only one case report of an infant about airway management of DS in literature [3]. We share our experience with an adult DS patient who underwent femoral derotation osteotomy under general anesthesia.

CASE PRESENTATION

A 20-year-old male patient with DS was consulted for femoral derotation osteotomy. The written consent has been obtained from the patient. The patient's height was 139 cm (<3 percentile), weight was 68.5 kg (50-75 percentile), and head circumference was 51.5 cm (<3 percentile. His body mass index was 36 kg/m^2. He had a short neck, prognathism, depressed nasal ridge (Figure 1). His Mallampati score was III. There were no other signs about a difficult airway at his physical exam. He was taken to the operating room with Covid-19 pandemic precautions.

Figure 1. 20-year-old male patient with Desbuquois Syndrome. His height was under 3 percentiles. He had a short neck, prognathism, depressed nasal ridge. His mallampati score was III.

After routine monitoring, the patient was pre-oxygenated with 100% FiO2 for 5 minutes. Rapid sequence induction was performed with propofol, fentanyl, and rocuronium. After induction, invasive artery and temperature monitoring was performed. Mask ventilation was easy. He was intubated using a 7.5 cuffed endotracheal tube with GlideScope® videolaryngoscope (GVL; Verathon, Bothell, WA, USA). His Cormack-Lehane score was II. Total intravenous anesthesia (propofol and remifentanil) was used for maintenance. After the operation was completed without any problems, muscle relaxation was reversed with sugammadex. He was extubated in the operating room and transferred to the post anesthesia care unit.

DISCUSSION

Desbuquois Syndrome is a rare syndrome with autosomal recessive osteochondral dysplasia [4]. Although it is mentioned that DS has a mortality rate of 33%, the exact mortality can't be calculated when it is considered that there are also milder forms that are not diagnosed [5]. Our patient was also a mild form of DS. The diagnosis of DS of our patient was made when the c375 G>C homozygous mutation was positive in the 2nd exon of the CANT1 gene.

Although there is a risk of a difficult airway, we could not apply regional anesthesia to the patient instead of general anesthesia because the operation time would be long. The use of videolaryngoscopy as the first choice was a factor that facilitated intubation especially in the presence of expected difficult intubation and Coronavirus pandemic precautions. Although inhalation anesthesia was mentioned in the previous case reports, we thought that total intravenous anesthesia was safer in terms of malignant hyperthermia due to musculoskeletal anomalies in DS. We preferred to use total intravenous anesthesia in our case. Total intravenous anesthesia can be used in these patients to avoid the risk of malignant hyperthermia, although it is not clearly mentioned in the literature. Some other anesthetic implications that may accompany DS like joint dislocations (including cervical vertebrae), gastroesophageal reflux, and respiratory and

cardiac involvement should be considered as well as airway management [6,7]. Because of the risk of joint deformity, we performed laryngoscopy gently and carefully. We think that our case will contribute to the literature in terms of being an airway management of an adult case. Adult patients with DS can be managed safely with adequate difficult airway preparation.

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The data of the study were presented for the first time as a poster presentation at the Balkan States Anesthesia Days - VII held on 30 April- 2 May 2021.

Author contribution

Study conception and design: MT, BB, and FS; data collection: HK and MT; analysis and interpretation of results: MT; draft manuscript preparation: AAY and MT. All authors reviewed the results and approved the final version of the manuscript.

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Conflict of interest

The authors declare that there is no conflict of interest.

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