Rhizomelic Chondrodysplasia Punctata: Respiratory Challenges

Sonia Mehta, MD; Joy Allee, MD;
Department of Anesthesiology, University of Florida College of Medicine, Gainesville, FL

Purpose

Rhizomelic chondrodysplasia punctata (RCDP) is a rare Congenital syndrome that is part of a subgroup of the larger classification, chondrodysplasia calcificans punctata (CCP). RCDP is manifested clinically by shortening of long bones, cataracts, malformation of facial features, failure to thrive, and severe mental retardation. These patients often have extremely low birth weight, body length, and head circumference. Diagnosis of this disease is made clinically through calcified stippling of the hyaline cartilage. Several case reports of RCDP have been written with anesthetic concerns focused mainly on tracheal stenosis. We present other respiratory challenges encountered on a 4-week-old patient with a diagnosis of RCDP, who presented for congenital cataract extraction.

Clinical Features

A 4-week-old, 2.7 kg patient, with a medical history of RCDP presents for a cataract removal. The patient had an uneventful G tube placement with anesthesia, at an outside institution. No perioperative complication was reported to the mother after the G tube procedure. Anesthesia induction was achieved with oxygen and sevoflurane via face mask, followed by establishment of intravenous (IV) access. Rocuronium 3 mg IV was then administered to facilitate the intubation process. Upon direct laryngoscopy, this patient was found to have a grade 3 view with deep cricoid pressure application. In between laryngoscopy attempts, it was necessary to maintain the patient on 100% fractional inspired oxygen level because he experienced significant desaturations, while also in the midst of challenging mask ventilation. After successful advancement of a 3.0 uncuffed endotracheal tube (ETT), breath sounds were absent in the left long fields, until the ETT was withdrawn to a depth of 7 cm at the lips. This raised a clinical suspicion of a shortened trachea. Following the operation the patient was successfully extubated, and monitored overnight at an inpatient unit.

Figure 1

Figure 2

Dysmorphic ears, coarse facial features, small lower jaw, and scanty hair are evident

Severe tracheobronchial stenosis in the X-linked recessive form of chondrodysplasia punctata

Discussion

It is essential that the anesthesiologist understands the respiratory challenges affiliated with RCDP patients, prior to inducing anesthesia. There are multiple case reports demonstrating the craniofacial abnormalities associated with RCDP. These craniofacial abnormalities have led to difficulties with mask ventilation and securing the airway on these patients. Although tracheal stenosis and respiratory tract cartilage changes have already been reported in the literature; our encounter with challenging mask ventilation, difficult laryngoscopy, and clinically relevant tracheal shortening, has yet to be described. It is suggested that a child weighing 2 kg should have a ETT depth of 9.6cm. Our team became suspicious of tracheal shortening when the ETT depth had to be withdrawn to 7 cm to appreciate bilateral breath sounds. With an ETT not long enough the cuff was partially protruding out from the vocal cords. This has the potential for complications that could lead to vocal cord dislocation, inadequate protection of the airway, or cord inflammation leading to postoperative airway complications. It is critically essential to be adequately prepared to handle the tenuous airway of RCDP patients. This preparation should span from a thorough preoperative clinic visit, to having a variety of difficult airway equipment and an experienced airway practitioner readily available.

References

2. Braverman NE. Rhizomelic Chondrodysplasia Punctata Type 1. 2001 Nov 16 [Update 2012 Sep 13].