A Rare Case of Pulmonary Hypoplasia and Complete Tracheal Rings in a patient with Cleft Lip and Palate

Faiz S. Nasser, MD; Jewel D. Montgomery, MD; Judith L. Ponder Handley, MD

Department of Pediatric Anesthesiology
University of Oklahoma College of Medicine, Oklahoma City, Oklahoma

INTRODUCTION

We present a case of intraoperative management of cleft lip repair leading to the diagnosis of cleft lip and palate with associated complete tracheal rings and unilateral pulmonary hypoplasia. Cleft lip and palate are two of the more common congenital malformations, with an estimated incidence of 1 in 700 births. Cleft lip repair is most often performed within the first 2-3 months of life, while cleft palate repair occurs at 6-9 months. Cleft lip and palate are associated with more than 300 syndromes. The most common syndrome involving cleft lip/palate, tracheal, and cardiovascular abnormalities is 22q11 deletion syndrome (DiGeorge or velo-cardio-facial syndrome). A case of isolated unilateral pulmonary hypoplasia with complete tracheal rings in a child with cleft lip and palate has not been described in the literature.

CASE PRESENTATION

A 2-month-old male with history of cleft lip and palate presented to the University of Oklahoma Children’s Hospital for cleft lip repair. Cleft lip and palate were identified by ultrasound at 17 weeks gestation to a healthy 33 year-old woman on her first pregnancy. The mother only took prenatal vitamins and denied history of smoking, alcohol or illicit drug use. The father had two healthy daughters by another union. She underwent unremarkable vaginal delivery with birth weight 6 pounds, 9 ounces. The neonate was discharged to home at 48 hours of life.

Preoperative history and physical were unremarkable and the patient classified as ASA I. He was brought into the OR and standard ASA monitors were placed and inhalational induction initiated with nitrous oxide and sevoflurane. Peripheral IV was placed and intubation was facilitated with IV propofol and fentanyl. Three attempts at direct laryngoscopy, first with cuffed 3.5 ET tube and subsequently uncuffed 3.0 ET tube failed despite grade I views. We were unable to advance the endotracheal tube past the cord. Mask ventilation was easy but asymmetric chest rise was observed with breath sounds heard only on the right. Intraoperative chest x-ray showed severe left-sided atelectasis.

A pediatric ENT surgeon performed a rigid bronchoscopy showing complete tracheal rings starting distal to the vocal cords with tracheal stenosis to the level just above the carina. Near complete left bronchomalacia with normal right bronchus was noted. We suspected that he may have an underlying cardiopulmonary abnormality and aborted the cleft lip repair. The patient tolerated the procedure well and was admitted for further workup.

He underwent a second anesthetic to obtain a CT scan of his neck and chest. It showed a hypoplastic left pulmonary artery resulting in hypoplasia of the left lung and with minimally aerated left lower lobe. Proximal tracheal narrowing was noted. Transthoracic echocardiogram showed severe hypoplasia of the left pulmonary artery, measuring 3.4 mm in diameter (by comparison, the right was 9.2 mm), but no significant gradient noted across either artery. Genetic testing was performed including CHD-7, FISH and DiGeorge Chromosomal Microarray. The results were normal with 46 XY karyotype and no interstitial deletions or duplications. The patient was discharged two days after admission. He subsequently underwent his cleft lip repair one month later with a 3.0 endotracheal tube placed under video bronchoscopy.

WORKUP

We present a rare case of complete tracheal rings with unilateral pulmonary hypoplasia in a patient with cleft lip and palate. To our knowledge, this is the first described case of these isolated anomalies in the literature. Cleft lip and palate repair are a commonly performed operation, yet there is no consensus on how these patients should be worked up, especially in asymptomatic patients such as ours. Cleft lip/palate have been associated with over 300 syndromes. One study identified cardiac malformations in 10% of 329 patients, including malformations of the septa, vasculature and valves. A distinction emerges between patients who present with cleft lip vs. those who present with cleft palate. One study analyzed 616 cleft infants (367 boys and 249 girls). 21% of these patients had associated malformations that required treatment or further follow-up.

CASE DISCUSSION

- Cleft lip - 8% malformations
- Cleft lip/palate - 28% malformations
- Cleft palate - 22% malformations

Cardiovascular malformations occurred 24% of the time, with congenital heart disease being most common. They noted that congenital heart disease was 16 times more likely to occur in a cleft palate patient compared to the general population and that routine ECHO screening may be justified.

Another study also found a difference between cleft lip compared to cleft lip.
- Cleft lip - 7.6-41.4% malformations
- Cleft lip/palate - 21.1-61.2% malformations
- Cleft palate - 22.2-78.3% malformations

The most common chromosomal abnormality was 22q11 deletion. Many of these patients had asymptomatic cardiac disease and were not diagnosed until later in life when speech and learning defects manifested. They recommended treating cleft lip and palate as two separate entities.

- If isolated cleft lip without any other known abnormalities, they did not recommend further testing.
- Patients with cleft palate, they recommended karyotype analysis.

Our patient underwent a total of three anesthetics. There is concern that early exposure to anesthetic agents may induce neurotoxicity and apoptosis. One study showed a 60% increased diagnosis of developmental and behavioral disorders among siblings who underwent an anesthetic before the age of 3 compared to those who did not. The relative risk increased with each exposure to an additional anesthetic. These studies are limited by their retrospective nature. The Pediatric Anesthesia Neuro-Developmental Assessment study is an ongoing cohort study comparing one sibling who received an anesthetic under 36 months of age vs. the other sibling who did not undergo surgery. The data suggests that preoperative testing of patients with cleft palate may be beneficial in accurately diagnosing patients prior to being put under an anesthetic. Given the increasing concern regarding anesthetic exposure in young patients, this may be a more prudent and safe course of action.

REFERENCES