Case Report

Successful anesthetic management of a child with blepharophimosis syndrome and atrial septal defect for reconstructive ocular surgery

Dalim Kumar Baidya, Puneet Khanna, Anil Kumar, Dilip Shende

Department of Anaesthesia and Intensive Care, All India Institute of Medical Sciences, New Delhi, India

Abstract

Blepharophimosis syndrome is an autosomal dominant disorder characterized by eyelid malformation, involvement of reproductive system and abnormal facial morphology leading to difficult airway. We report a rare association of blepharophimosis syndrome and atrial septal defect in a 10-year-old girl who came for reconstruction surgery of eyelid. The child had dyspnea on exertion. Atrial septal defect was identified preoperatively by clinical examination and echocardiography. Anesthesia management was complicated by failure in laryngeal mask airway placement and Cobra perilaryngeal airway was subsequently used.

Key words: Anesthesia, atrial septal defect, blepharophimosis syndrome, difficult airway

Introduction

Blepharophimosis syndrome is an autosomal dominant disorder due to defects in FOXL2 gene on chromosome 3q and predominantly affects eyelids. This syndrome is characterized by blepharophimosis, ptosis, and epicanthus inversus (BPES).[1] BPES can be associated with other systemic abnormalities and difficulty in airway management due to abnormal facial morphology. We report anesthetic management of a rare case of blepharophimosis syndrome associated with atrial septal defect (ASD) for plastic reconstruction of eyelids and review the relevant literature. There is no report in the literature so far on anesthetic management of blepharophimosis syndrome.

Case Report

A 10-year-old, 22 kg, girl presented with drooping of both upper eyelids since birth along with bilateral narrow horizontal palpebral fissure and skin tags of both lower eyelids over the medial canthus. On ophthalmic evaluation, the child was diagnosed to have blepharophimosis syndrome and was posted for sequential bilateral sling surgery.

There was history of mild breathing discomfort on climbing two flights of stairs for last 1 year. She denied any history of palpitation, chest pain, syncope, or swelling of feet. She had normal developmental milestones. On general examination, the girl maintained a chin up and head tilt backward posture along with raised eyebrows to clear the visual axis. The nasal bridge was flat. Her pulse rate was 90/min and blood pressure was 100/65 mmHg. There was no pallor, edema, or cyanosis. On systemic evaluation, an ejection systolic murmur, most prominent over the pulmonary area, was auscultated over the precordium. Chest X ray did not show any abnormal finding and electrocardiogram (ECG) revealed right axis deviation and incomplete right bundle branch block (RBBB). Echocardiography revealed 5 mm ostium secundum ASD with a left to right shunt. Biventricular function was normal. Hematological and biochemical investigations were normal. Airway examination revealed small mouth opening and high arched palate.

The child was premedicated with oral midazolam 0.5 mg/kg 45 minute before surgery. In the operating room, routine monitoring included pulse oximetry, ECG, noninvasive blood pressure, and end-tidal carbon dioxide and gas monitoring. Intravenous access was obtained and radial artery cannula was inserted for invasive blood pressure monitoring. Anesthesia

Address for correspondence: Dr. Dalim Kumar Baidya, Department of Anesthesia and Intensive Care, All India Institute of Medical Sciences, 8/67, Gent's Hostel, New Delhi - 110 029, India.
E-mail: dalimkumarb001@yahoo.co.in

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was induced with oxygen, sevoflurane and fentanyl 40 μg. Atracurium 10 mg was injected for muscle relaxation after ability to mask-ventilate the patient was confirmed. Insertion of proseal laryngeal mask airway (LMA) size 2.5 was unsuccessful despite two attempts (classical technique and direct-laryngoscopy guided technique consecutively). Direct laryngoscopy revealed a Cormach-Lehane grade IV. However, mask ventilation was adequate and subsequently Cobra perilaryngeal airway (CobraPLA) #2 was successfully inserted. Anesthesia was maintained with oxygen, air and sevoflurane with intermittent dose of atracurium and fentanyl. Pressure-controlled ventilation was used and normocarbia maintained. The surgery lasted for 75 minutes. The patient was hemodynamically stable throughout the intraoperative period. Ringer's lactate 500 ml was infused. At the end of surgery, neuromuscular blockade was reversed with neostigmine and glycopyrrolate and the CobraPLA was removed. There was minimal blood stain on the cuff of the CobraPLA. She was shifted to postanesthesia care unit and monitored for 2 hours. Postoperative analgesia was provided with paracetamol 500mg intravenously q6hr. Postoperative course was uneventful.

She was rescheduled for surgery of the other eye after 7 days. Airway was secured by CobraPLA #2 in the first attempt. Rest of the anesthetic management was similar and uneventful. Arterial cannula was not used at that time as the clinical course of the patient was stable and we expected minimal hemodynamic variation based on our previous experience.

She was discharged home 4 days after the second surgery and advised follow-up in the cardiology department for management of the ASD.

Discussion

BPES was initially described by Komoto in 1921.[2] BPES can be categorized into two types. Type I is characterized by the ocular finding associated with premature ovarian failure and infertility in females, whereas in type II only ocular involvement is found.[3] In type I BPES, the females have normal fertility in the early reproductive period, followed by oligomenorrhea and premature menopause before the age of 40. Ultrasound shows small hypoplastic uterus and streak ovaries. Endocrinological profile shows hypergonadotrophic hypogonadism with elevated follicular stimulating hormone, luteinizing hormone, and decreased serum concentration of estradiol and progesterone. Most of the affected females require early hormone replacement therapy.[4,5] Involvement of other systems in BPES is rare. Ocular and airway involvement similar to that of blepharophimosis syndrome can occur in Freeman-Sheldon syndrome, Dubowitz syndrome, and Noonan syndrome. These syndromes can have other systemic involvement along with blepharophimosis and airway involvement [Table 1].[6-10]

Patients with BPES syndrome can come for ophthalmic surgery in their childhood or later in adulthood. Females with infertility can come for diagnostic laparoscopy or *in vitro* fertilization. This patient had not yet attained menarche. Endocrine and gynecology evaluation was done and preoperative ultrasound and hormone profile were considered unnecessary. The relatives were informed about the possibility of infertility and advised follow up in the gynecology department after discharge.

Difficult airway was anticipated due to the presence of flat nasal bridge, small mouth opening, and high arched palate. Muscle relaxant was injected after ensuring that mask ventilation was not difficult. In the presence of high arched palate, LMA insertion using classical technique may fail and laryngoscopy-guided LMA placement can be an alternative.[11] However, in the present case, due to a small mouth opening LMA insertion was unsuccessful. CobraPLA

Table 1: Anesthetic concerns in BPES and other related syndromes

a) BPES	
Airway involvement:[9]	
Flat nasal bridge	Difficult mask ventilation
Small oral cavity	Difficult intubation/LMA placement
High arched palate	Difficult intubation/LMA placement
Amblyopia and strabismus	Pre-operative documentation of vision (particularly for non-ophthalmic surgeries)
Systemic involvement:	
Hypergonadotrophic hypogonadism	Infertility, early hormone replacement therapy
Mental retardation, developmental abnormality[10]	Very rare
b)Freeman-Sheldon syndrome[6]	Blepharophimosis, hypertelorism, a flat nose, microstomia, micrognathia, scoliosis and multiple arthrogryposis
c) Dubowitz syndrome[7]	Blepharophimosis, bilateral ptosis, a flat nasal bridge with a broad nasal root and micrognathia. cleft palate, tooth problems and cranio-cervical anomalies
d) Noonan syndrome[8]	Ptosis, epicanthus, hypertelorism, high arched palate, short stature, mental retardation, congenital heart disease, scoliosis, pectus carinatum or excavatum

BPES: Plepharophimosis, ptosis, and epicanthus inversus

insertion has been found to be more successful in limited mouth opening and it provides better airway seal and a conduit for intubation.[12] Use of CobraPLA has been reported as a rescue airway device *in situ*ations where intubation and LMA insertion failed.[13]

Congenital heart disease (e.g., ventricular septal defect and complex heart defect) have been reported to be associated with blepharophimosis syndrome.[14] Clinical history of exertional dyspnea and presence of ejection systolic murmur in this patient led us to suspect congenital heart disease. Echocardiography confirmed the diagnosis of ASD. But there was no significant chamber enlargement or ventricular dysfunction. The presence of ASD did not pose any significant hemodynamic problem. Premedication was provided to ensure a calm child during induction. Infective endocarditis prophylaxis, however, was not considered.[15] Anesthetic drugs and techniques used were targeted to prevent any sudden or significant change in systemic and pulmonary vascular resistance. Arterial cannula was obtained to detect minimum and beat to beat fluctuations of blood pressure and for better titration of anesthetic agents. All precautions were obtained to prevent air bubble injection in the intravenous cannula. Postoperatively, the child was kept on a monitored bed for the first 24 hours.

In conclusion, blepharophimosis, ptosis, and epicanthus inversus syndrome can be associated with systemic involvement, difficult airway, and rarely with congenital heart disease posing challenges in anesthetic management. High index of suspicion should be maintained to appropriately diagnose the associated anomalies and anesthetic management should be tailored accordingly.

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