Case Report

Anesthetic Management of a Child with Diamond-Blackfan Syndrome

Alok Narasimha, Nischala Dixit, Latha P John, Swaraj Cherian

Department of Anaesthesiology, St. John's Medical College and Hospital, Bengaluru, Karnataka, India

Abstract

Diamond-Blackfan anemia (DBA) is a rare congenital disorder that can pose a variety of anesthetic challenges to a clinician. A disorder of pure red cell aplasia is associated with other congenital anomalies such as craniofacial malformations, thumb or upper limb abnormalities, cardiac defects, urogenital malformations, and cleft palate. These anomalies are compounded by problems of iron overload and chronic steroid therapy. Anesthetic management of a child with DBA requires knowledge and skill to handle a difficult pediatric airway and a thorough understanding of the congenital heart lesion of the child and its interaction with anesthetic agents and surgery. Rarity of DBA, along with its associated problems and lack of awareness of these by the anesthesiologist, makes the occasional surgery in this population very challenging. We report a 1-year 3-month-old child, diagnosed to have DBA during infancy, posted for laparoscopic orchidopexy.

Key words: Anemia, Diamond-Blackfan, general anesthesia, pure red cell aplasia

Introduction

Diamond-Blackfan anemia (DBA) is a syndrome with inherited erythroblastopenia[1] and has a prevalence of 5-7 per million live births.[2] Anemia is diagnosed early in life, usually before the age of 1 year with a median age of diagnosis at 3 months. The principal warning signs are pallor and dyspnea, especially during feeding or suckling, failure to thrive, and systolic murmur. Patients have unambiguous red cell failure, defined by macrocytic anemia and reticulocytopenia and decreased or absent red cell precursors in the bone marrow.[3] DBA is an inherited disorder with mutation on the gene localized to chromosome 19q13.3. Therapeutic approaches include regular transfusions and long-term administration of corticosteroids. Radical treatment for this disorder is allogenic bone marrow transplantation. Affected children have a variety of congenital abnormalities including craniofacial malformations, thumb or upper limb anomalies, cardiac defects, urogenital malformations, and cleft palate. Life expectancy is one-two decades.

Case Report

A 1-year 3-month-old child weighing 7 kg presented to the pediatric surgery department with empty scrotal sac noticed by the parents since birth, posted for laparoscopic orchidopexy. At the age of 1 month, the child presented with progressive pallor, excessive lethargy, and one episode of unresponsiveness. On examination, the child had a large dysmorphic head, microstomia, large tongue, retrognathia, short neck, and other features such as right ear microtia, hypoplastic right thumb, hypospadias, and undescended testis. There was no hepatosplenomegaly. Bone marrow examination showed cellular marrow with erythroid hypoplasia. Echocardiogram revealed atrial septal defect and coronary cameral fistula draining into the right ventricle (RV). Abdominal ultrasonography (USG) showed ectopic right kidney with suspicious intraabdominal left testis. The child had multiple blood transfusions in view of anemia and was started on oral administration of prednisolone of dosage 5 mg three times a day (TID) from the age of 1 year. Hemoglobin estimated preoperatively was 9.1 g% following treatment.

After a preanesthetic evaluation, high-risk consent was obtained and a bed in the pediatric intensive care unit was arranged for postoperative ventilation if required. On the

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Address for correspondence: Dr. Alok Narasimha, Department of Anaesthesiology, St. John's Medical College and Hospital, Sarjapur Road, Bengaluru - 560 034, Karnataka, India. E-mail: aloknarasimha@gmail.com

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day of surgery, a difficult airway cart including supraglottic airway device, fiber optic bronchoscope of size 2.8 OD, and gum elastic bougie were kept ready. In view of the difficult airway, gaseous induction was carried out with oxygen and titrated doses of sevoflurane. Intraoperative monitors included echocardiogram (ECG), pulse oximetry, and capnography. After having secured an intravenous access, 0.05 mg of glycopyrrolate and 15 µg of fentanyl were given. Laryngoscopy that was carried out revealed glottic view of Cormack and Lehane's grade III. With external laryngeal manipulation, a 4-mm internal diameter (ID) uncuffed endotracheal tube was inserted using a stylet. After confirming endotracheal tube placement, atracurium of dosage 5 mg was administered. Then, 200 mL of Ringer's lactate was administered for fluid management. Anesthesia was maintained with oxygen, air, and isoflurane with intermittent doses of fentanyl and atracurium. The surgery lasted for 1 h following which residual neuromuscular blockade was reversed and the child was extubated when fully awake. The child was shifted to the pediatric post anesthesia care unit for further management [Figure 1].

Discussion

First noted by Joseph in 1936, the condition is, however, named after Diamond and Blackfan, who described congenital hypoplastic anemia in 1938.[3]

The initial clinical manifestations are those of an isolated anemia during infancy:[4] Pallor, shortness of breath, failure to thrive, and a systolic murmur. Anemia can be moderate or severe. It is usually macrocytic with persistently low reticulocyte counts. The sizes of the liver and spleen are normal.

Diagnosis is primarily by blood count and bone marrow biopsy, which shows the absence or rarity of erythroid precursors that have a normal morphological appearance. This is supported by the presence of elevated fetal hemoglobin, elevated adenosine deaminase levels in red blood cells, and congenital abnormalities.[4]

Among DBA-affected children, 10-40% also have craniofacial malformations, thumb or upper limb abnormalities, cardiac defects, urogenital malformations, and cleft palate. They are at a modest risk of developing leukaemia and other malignancies. Additional problems are due to long-term steroid therapy and iron overload secondary to multiple blood transfusion. Significant accumulation of iron in tissues may occur within a few years in transfusion-dependent patients. Clinical manifestations depend on the affected organ and may include heart failure/cardiomyopathy, diabetes, arthritis, cirrhosis, and liver cancer. Children at risk should be screened for iron overload.[5]

Anesthetic considerations would be an abnormal airway anatomy, congenital cardiac defects, and anemia, along with implications of a child on exogenous corticosteroids. A child with DBA posted for surgery under anesthesia should have a detailed examination of the airway, evaluation of the possible congenital heart defects by echocardiography, perioperative replacement of corticosteroids, and optimization of hemoglobin preoperatively.

The anticipation of a difficult airway and planning and preparation for the same ensure optimal management. The advantages of general anesthesia should be balanced against the potential loss of airway protective reflexes and spontaneous ventilation.[6] Use of intravenous induction agents, along with neuromuscular blockade, results in loss of protective airway reflexes that is undesirable. Sevoflurane is the induction agent of choice in pediatric anesthesia, isoflurane and desflurane being used for maintenance.[7] An intermediate acting nondepolarizing muscle relaxant can be used after securing the airway. Analgesia can be maintained by titrated doses of opioids, port site injection of local anesthetics in laparoscopic surgeries, rectal paracetamol suppositories, and caudal epidural injection of local anesthetics in the absence of anogenital and spine anomalies.[8]

Congenital heart disease (CHD) associated with DBA needs attention during anesthetic management. Information about the cardiac lesion, its altered physiology, and its implications under anesthesia should be obtained. Neonates and infants with CHD experience a twofold increase in mortality from noncardiac surgery. Inhalational induction is acceptable in CHD patients with uncomplicated cardiac lesion. 100% oxygen and hyperventilation in patients with left-to-right (L-R) shunt will result in pulmonary vasodilation that in turn increases pulmonary congestion and should thus, be avoided. High pulmonary flow in unrestricted L-R shunt [large atrial septal defect (ASD)] will lead to congestive heart failure (CHF) and pulmonary hypertension (HTN).[9] Extreme vigilance and air

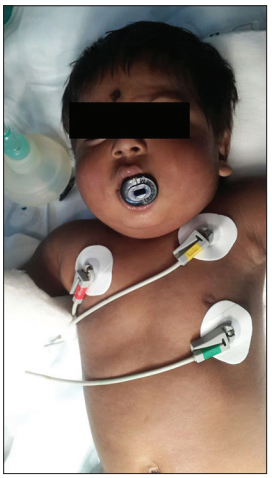

Figure 1: Child with Diamond Blackfan syndrome

traps are vital to prevent air embolism in cases of ASD during intravenous injections.

The child may be extubated following surgery after complete reversal of the neuromuscular blockade and observation in post-anesthesia care unit (PACU).

Katircioglu *et al.,* managed a child aged 11 years for open reduction and internal fixation of femur fracture under general anesthesia. The child was on transfusion therapy with prolonged high dose of steroids for her anemia. Katircioglu *et al.,* did not encounter any difficult airway. Corticosteroid was administered preoperatively.

Conclusion

Multiple anomalies and anemia can be challenging for anesthesiologists in managing patients with DBA. Anesthetic management should be based on a through preoperative assessment of airway cardiac, musculoskeletal, hematology, and craniofacial abnormalities. Perioperative steroid supplementation should be considered if these children are on prolonged steroid therapy.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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