Case Report

Ocular surgery in a child with Coffin Lowry syndrome: Anesthetic concerns

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Abstract

Coffin Lowry syndrome is a rare disease involving multiple organ systems. From the anesthesiologists point of view it involves mental retardation, seizures, difficult airway, cardiac abnormalities (pediatric dilated cardiomyopathy) and skeletal deformities. We share our experience of management of a child with Coffin Lowry syndrome and also discuss the problems faced during perioperative period.

Key words: Coffin Lowry syndrome, difficult airway pediatrics, dilated cardiomyopathy pediatrics

Introduction

Coffin Lowry syndrome is a rare disease involving multiple organ systems. Commonly it presents in males with mental retardation, facial abnormalities, cardiovascular involvement, and skeletal deformities. An anesthesiologist faces many unique challenges due to multi-system involvement and the problem is compounded by limited literature on this topic. We share our experience of successful management of a child with Coffin Lowry syndrome for ocular surgery. Most of the associations of this syndrome may not have complete penetrance and may be clinically absent. However, in the case presented, all systemic ailments were present simultaneously which added to the complexity of anesthetic management.

Case Report

A 25-kg, 14-year-old boy, a known case of Coffin Lowry syndrome, presented with high myopia and retinal detachment associated vision deterioration. Vitreo-retinal surgery was scheduled. At 2 years of age, he was diagnosed with Coffin Lowry syndrome which was confirmed by genetic analysis. He was born at term without any history of birth asphyxia. Since the age of 3 months, he had been suffering from recurrent chest infections requiring frequent hospital admissions and occasional mechanical ventilation. Severe mitral regurgitation was also diagnosed, and by 3 years, he developed dilated cardiomyopathy (DCM) [Figure 1] with heart failure. Patient was on oral furosemide and digoxin therapy for last 10 years but had no sign of failure at presentation. Patient developed hypothyroidism at infancy and received thyroxine for 13 years. The thyroxine had been tapered off 1 year back, and patient was clinically and biochemically euthyroid since then. At the age of 4 months, he developed recurrent myoclonic seizures for which combination of multiple anti-epileptics was administered, but they were stopped after he remained asymptomatic for a 5-year period. He complained of severe reflux gastritis and vomiting a year back for which daily ranitidine and metoclopramide were prescribed. With growth, he developed scoliosis. It was not possible to communicate with him directly due to his mental status; however, he seemed to reply appropriately to his father. Since Coffin Lowry syndrome is an X-linked dominant trait, we tried to explore for similar family history, but nobody from the maternal or paternal lineage was found to possess any clinical manifestations.

On examination, an abnormal facies was evident with thick everted lips, increased intercanthal distance, and frontal bossing. Scoliosis and pectus carinatum deformity were present. Pitting pedal edema till ankle was present in both the feet. On auscultation, a crescendo murmur was heard in cardiac apical region. The air entry was normal in both lung fields and there were no added sounds.

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Access this article online	
Quick Response Code:	Website: www.joacp.org
	DOI: 10.4103/0970-9185.105818

The airway was Modified Mallampati Class III, and there was a large tongue, high-arched palate, receding mandible, and buckteeth. Mouth opening and neck movements were within normal ranges. Hemogram, liver, renal, and thyroid function tests were normal. Echocardiography revealed mild mitral regurgitation (MR) with thickened mitral leaflets and severe pulmonary artery hypertension (PAH) with mean pressure of 80 mmHg, which could not alone be attributed to the degree of MR seen on 2D echocardiography. A Cobb's angle of 42° without any cord involvement was seen on magnetic resonance image spine. Room air oxygen saturation was 99% with pulse rate of 102/min.

Written informed paternal consent was obtained and the child was allowed water till 2 h prior to surgery. In view of history of heart failure, a conservative approach toward preoperative fluids was adopted and no fluids were administered during the fasting period. Patient was advised to continue ranitidine, metoclopramide, furosemide, and digoxin. After ensuring normal electrolyte values in the morning, oral midazolam syrup 12 mg was given half an hour prior to shifting to operating room.

Patient was shifted to the operating room, along with his father to keep him calm. After connecting standard monitoring, anesthesia was induced inhalationally with sevoflurane in 100% oxygen. While maintaining his spontaneous ventilation, intravenous (IV) access was obtained and left radial artery was cannulated with a 22-G cannula. Fentanyl 50 mcg and atracurium 10 mg were administered IV after which the child was hand ventilated ensuring low inspiratory pressure. No Positive End Expiratory Pressure (PEEP)was used. Confirming adequate depth of anesthesia, a 2.5 size proseal laryngeal mask airway (LMA) was inserted and its placement confirmed. Pressure-controlled ventilation without PEEP was used avoiding hypercarbia throughout the procedure. Anesthesia was maintained on oxygen, air, and sevoflurane. During surgery (duration 70 min), he received 100 ml of Ringer lactate and another 20 mcg of fentanyl. At the end of surgery, neuromuscular blockade was reversed, and once the patient was fully awake, the LMA was removed. Patient was shifted to the intensive care unit (ICU) for postoperative monitoring. The postoperative course was uneventful. Ketorolac was administered IV for analgesia and fentanyl IV was used as rescue analgesic. Patient was shifted to the ward the next day.

Discussion

Coffin Lowry syndrome is a rare disease which presents clinically as hypotonia and lax joints at birth. As the child grows, facial and airway abnormalities and other systemic manifestations begin to appear. Common associated abnormalities of anesthetic importance are enumerated in Table 1 and[1] Figure 2. Preoperative assessment of such

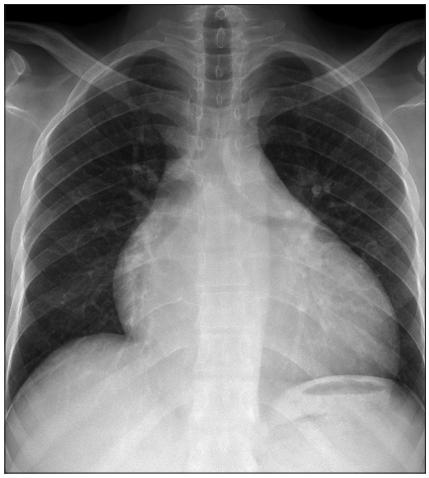

Figure 1: Chest X-ray showing dilated cardiomyopathy

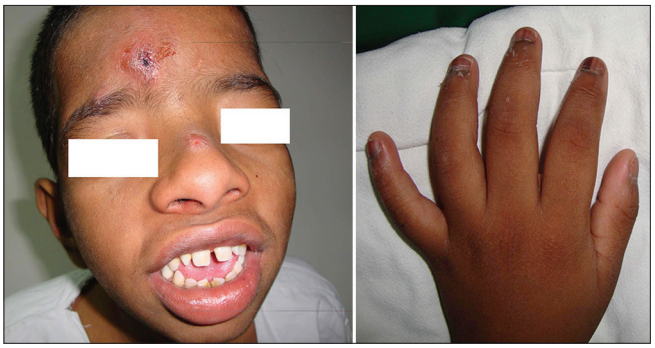

Figure 2: Typical facies and short fleshy fingers

Table 1: Anomolies seen in Coffin Lowry syndrome

Facial profile	Macrocephaly
	Frontal bossing
	Maxillary hypoplasia
	High-arched narrow palate
Airway	Large tongue, large mouth
	Thick everted lips
	Broad nose
	Hypodontia
	Peg-shaped incisors
Cardiac	Mitral valve disorders
	Dilated cardiomyopathy
	Heart failure
	Pulmonary hypertension
Neurological	Mental retardation
	Seizures
	Stimulus-induced drop attacks
	Hypotonia
Skeletal	Kyphoscoliosis
	Pectus carinatum/excavatum
	Short stature
	Short stout limbs/fleshy fingers

mentally retarded children is difficult, thus parental presence and involvement is of utmost importance for assessment of growth history, natural history of the disease, and its current status.[2]

Cardiac failure with dilated cardiomyopathy is rare in the pediatric age group. Mitral valve disease with associated cardiomyopathy is known in this syndrome,[1,3] making echocardiography evaluation mandatory. Patients should be medically optimized on diuretics and digoxin. Diuretics can cause dehydration and electrolyte disturbances. Hypokalemia can precipitate digoxin-associated toxicity, and therefore preoperative electrolyte imbalance must be corrected.[4] Digoxin slows heart rate, increases inotropy, and may increase the regurgitant fraction in MR. Increase in peripheral resistance should be avoided as this can decrease systemic blood flow.[5] This increase in regurgitant fraction can potentially cause heart failure. In case of bradycardia and hypotension, atropine or glycopyrrolate is preferred over drugs like ephedrine or mephentramine which cause peripheral vasoconstriction and increase regurgitation. We tried to maintain the heart rate around the preoperative value (110/min).

Severe PAH in this patient may be attributable to a combination of scoliosis and moderate MR. Nitrous oxide and PEEP were avoided as both are known to increase pulmonary vascular resistance.[6] Invasive arterial monitoring should be considered in such cases. Pre-induction arterial cannulation is difficult in children, and hence we preffered cannulating left radial artery after induction.

This patient had high-arched palate and receding mandible, both of which are predictors of difficult airway. We preferred inhalational induction of anesthesia and muscle relaxant was used only after confirming mask ventilation. Even though a flexible LMA is preferred for ophthalmological surgeries, due to history of severe reflux, a proseal LMA was used, keeping a fiber-optic bronchoscope standby. Chances of failed LMA insertion are less likely as most of the documented anomalies in this syndrome are limited to facial anatomy with no documented laryngeal anatomical abnormalities.[7]

Good analgesia prevents any sympathetic stimulation and rise in peripheral resistance and myocardial stress; hence, fentanyl and ketorolac were used. Fluid maintenance in such patients with heart failure, DCM, and MR is challenging. We avoided liberal fluid administration as it can precipitate an increase in MR and cardiac decompensation. Maintaining an optimal preload is important for adequate cardiac output. Oral fluids should be started as soon as possible, in the postoperative period, as fluid management becomes easier and safer.

Our patient had history of seizures, but was asymptomatic and was not on anti-epileptic therapy at the time of presentation. Coffin Lowry syndrome patients can be on multiple anti-epileptics (enzyme inducers/inhibitors), which can alter anesthetic requirements.[8] Our patient had history of hypothyroidism, but was biochemically euthyroid at the time of presentation. Literature does not document any association of hypothyroidism with the syndrome and it is difficult to comment whether these patients should be subjected to thyroid function tests routinely.

Postoperative management in ICU ensures a safe postoperative course as these patients have multiple system involvement. Utmost caution is needed while sedating these children in the ICU as hypercarbia can worsen PAH.

To conclude, careful preoperative cardiovascular and neurological assessment, proper airway management, knowledge of different systemic involvement and drugs used, and low threshold for invasive monitoring ensure successful perioperative outcome of children with Coffin Lowry syndrome.

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How to cite this article: Singh PM, Baidya DK, Govindarajan S, Trikha A. Ocular surgery in a child with Coffin Lowry syndrome: Anesthetic concerns. J Anaesthesiol Clin Pharmacol 2013;29:114-6.
Source of Support: Nil, **Conflict of Interest:** None declared.