Case report

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Anaesthetic management of a patient with Cornelia De Lange syndrome

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ABSTRACT

Cornelia De Lange syndrome (CdLS) is a very rare genetic disorder. Anaesthesiologists facing such type of syndromic patients is also rare and management of these patients with mental and growth retardation has always been challenging. We present successful anesthetic management in a child with CdLS posted for cataract surgery.

Keywords: Anaesthesia, Cornelia De Lange Syndrome, Cataract

INTRODUCTION

Cornelia de Lange syndrome (Amsterdam dwarfism) a rare malformation syndrome, was first described in 1933 by a Dutch pediatrician Dr. Cornelia de Lange, after whom the disorder has been named[1,2]. Features of this syndrome include mental retardation, behavioral problem, short stature, limb abnormalities (mainly upper extremities) and distinctive facial features[1,2]. Many affected individuals also have behavior problems similar to autism. Other signs and symptoms of Cornelia de Lange syndrome (CdLS) can include excessive body hair (hypertrichosis), small head (microcephaly), hearing loss, genitourinary defect and problems with digestive tract[2]. Some people with this condition are born with cleft lip/palate, seizures, heart defects and eye problems.

CdLS is a very rare genetic disorder. Anaesthesiologists facing such type of syndromic patients is also rare and management of these patients with mental and growth retardation has always been challenging. We present successful anesthetic management in a child with CdLS posted for cataract surgery.

CASE REPORT

A 14-year male child, weighing 35 kg, who was a diagnosed case of CdLS, operated case of cleft lip and palate at the age of 3 year, was planned for cataract surgery. Pre-operative examination of child showed microbrachycephaly, bushy eye brow, micro-ophthalmia micro-cornea with coloboma in both eyes and congenital cataract in both eyes, depressed nasal bridge with anteverted nare, long philtrum, operated cleft palate and lip, protruded and widely spaced central incisors, macroglossia, high arch palate and short neck. Musculoskeletal examination revealed kyphosis, wide spaced nipple, short upper limb with syndactyly of 2nd and 3rd finger with simian crease. Patient had mild growth and mental retardation with speech impairment.

Routine biochemical investigations were within normal limits. Chest X-ray PA view showed no abnormalities. ECG and echocardiograbhy were also within normal limit.

A written informed consent was taken and the patient was kept nil per oral for six hours prior to surgery. Difficult airway was anticipated due to microcephaly, high arch palate and macroglossia. Based on medical history, and considering previous CdLS case reports, a basic anesthetic plan was made. Difficult airway kit including variable sizes of facemasks, oral airways, LMA and emergency cricothyroidotomy kit were kept ready. On arrival of the patient in the operation theatre, baseline ECG, pulse oximetry (SpO_2) and Non-Invasive

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Blood Pressure (NIBP) was recorded using multipara monitor (Philips MP30). Peripheral intravenous access with 22G iv cannula was secured on the dorsum of left hand. The patient was premedicated for aspiration prophylaxis with ondansetron 0.1 mg/kg and ranitidine 1 mg/kg intravenously. In addition, inj glycopyrolate 0.2 mg, inj midazolam 0.05 mg/kg, inj pentazocin 0.6 mg/kg intravenously was administered. We planned general anaesthesia with LMA insertion for cataract surgery. Patient was preoxygenated with 100% oxygen for 3 min before induction which was done with inj propofol 2.5mg/kg iv, and sevoflurane (6–8%). LMA size 3 was inserted and inflated with 20 ml air. Anesthesia was maintained on 100% O_2 and sevoflurane (2–3%) and on spontaneous respiration Hemodynamic and other vital parameters were stable throughout the entire duration of surgery (30 min). LMA was removed after proper suction of oral cavity and return of reflexes (Figure 1–4).

Figure 1

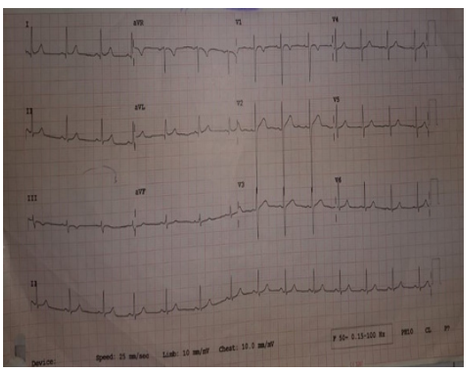

Figure 2

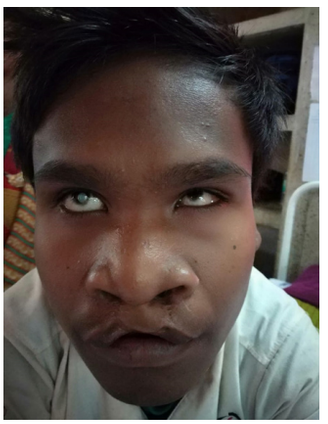

Figure 3

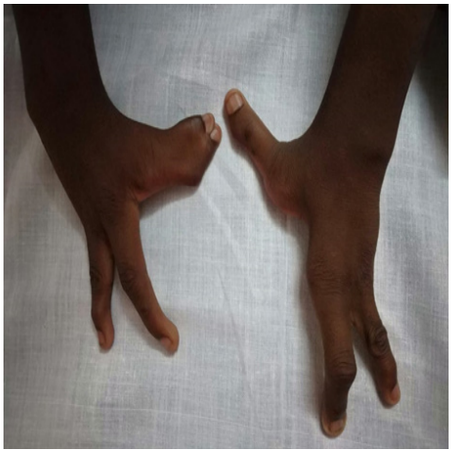

Figure 4

DISCUSSION

The Cornelia de Lange syndrome (CdLS) or Brachmann de Lange Syndrome is a rare congenital anomaly syndrome. The possibility of diagnosing this syndrome at birth is 1 case per 10,000 to 50,000 births[3]. Mutation in five genes (NIPBL, SMC1A, SMC3, RAD21, and HDAC8) has been identified[3]. Approximately 60% of CdLS are due to NIPBL mutation, 5% caused by mutation in SMC1A, RAD21 and HDAC8 and 5% caused by SMC3 mutation[2-3]. This syndrome is characterized by typical facial dysmorphism, growth and mental retardation, distinct craniofacial feature (including brachycephaly, hypoplastic mandible, cleft palate, macroglossia, upward facing nostrils and thin lip), ocular defects, epilepsy and varying degree of hirsutism[1,2].

Common ocular disorders associated with CdLS are long eye lashes and eyebrows that meet at center (synophrys)[8]. Synophrysis found in 99% of people with CdLS[1-3]. Most common findings include conjunctivitis, blepharitis, microcornea and corectopia. Aniridia and congenital glaucoma are also reported[8,9]. In our case we found synophrys, microophthalmia, microcornea with coloboma iris and congenital cataract in both eyes.

Anesthesiologists encountering such patients are rare. The various challenges for anaesthesiologists include difficult airway, hyper active airway, risk of aspiration, convulsion, cardiac arrhythmias, behavioral disturbance, prolonged apnea and malignant hyperthermia with use of suxamethonium, and prolonged awakening, recurrent respiratory infection with compromised lung function due to severe scoliosis and other musculoskeletal anomalies[4,5]. The craniofacial features associated with difficult airway include microcephaly, macroglossia with wide spaced prominent central incisors, high arched palate and short stiff extended neck.

Most of the documented characteristic features of CdLS were found in our case. Also, biochemical investigations were

within normal limits. Congenital heart anomalies such as ASD, VSD, Patent ductus arteriosus, hypoplasia of left ventricle and anomalies in ECG are often reported with this syndrome[6]. In our case we found normal ECG and ECHO finding.

Recurrent respiratory infection due to low immunity and pulmonary hypoplasia or lobular anomalies has been reported[5,6]. Our patient posed the danger of respiratory compromise and other post-operative pulmonary complication as he had common cold and kyphosis. Airway is considered to be irritable, as a patient with this syndrome is very sensitive to respiratory tract infection. Post-operative pulmonary complications are more common in such patients due to recurrent respiratory infections, irritable airway and more episodes of hypercapnia and hypoxia[5-7].

We avoided nitrous oxide because of the risk for increased pulmonary vascular resistance. Delayed recovery from general anesthesia in such patients has been mentioned in some cases in spite of use of sevoflurane as inhalational agent and atracurium, a short acting neuromuscular relaxant[4]. But fortunately there was no delayed recovery in our case inspite of using sevoflurane, the duration of the surgery was also kept short (30 min).

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