CASE REPORTS

ANAESTHETIC MANAGEMENT OF A PATIENT WITH CORNELIA DE LANGE SYNDROME

Ertan ÖZTÜRK, M.D., Füsun BOZKIRLI, M.D., Zerrin ÖZKÖSE, M.D.

Gazi University Faculty of Medicine, Department of Anaesthesiology and Reanimation, Ankara-Turkey
Gazi Medical Journal 2000; 11: 175-177

ABSTRACT : *Patients with Cornelia de Lange syndrome may encounter potential anaesthetic problems like seizures, cardiac abnormalities, and difficult tracheal intubation. This report describes the management of a 9-year-old girl with Cornelia de Lange syndrome who underwent thyroductal cyst excision with total intravenous anaesthesia, and reviews the literature on this syndrome and anaesthetic problems.*

Key Words: *Cornelia de Lange Syndrome, Intravenous Anaesthesia, Propofol.*

INTRODUCTION

Cornelia de Lange syndrome, first identified in 1933 by Cornelia de Lange, is a rare condition of mental retardation with a distinctive face, retarded growth and skeletal abnormalities (1, 2). Although a genetic aetiology has been proposed with suggestions of autosomal dominant and recessive inheritance, its aetiology is still unknown (3). It affects one in 30,000 to one in 60,000 live births (3, 4). Two thirds of the patients die before the end of their first year (1). Death occurs from pulmonary aspiration in infancy, and from infections and bowel obstruction in later life (1,3). The first reference in the literature to the anaesthetic management of the syndrome highlighted potential problems with seizures, cardiac abnormalities and difficult tracheal intubation (5). It is almost impossible to state the exact number of cases already on record because of the numerous publications from many countries. But we have found only ten reported cases about the anaesthetic management of Cornelia de Lange syndrome through Medline (up to August 2000).

CASE REPORT

A 9-year-old girl with Cornelia de Lange syndrome, treated three times because of infected thyroglossal duct cyst during last year, was scheduled for a surgical excision. Cornelia de Lange syndrome was diagnosed at the age of three when she failed to progress. She has one healthy sister and a twin sister with Cornelia de Lange syndrome. She was mentally retarded and her speech was abnormal. Her height (108 cm) and weight (16.5 kg) were below the third percentile. She had characteristic facial features, with microcephaly and a prominent forehead. She

strabismus and low-set ears. The lips were thin and the mouth was small with a high-arched palate. She was unable to walk because of tight Achilles tendons and abnormal muscle tone. Chest X ray and laboratory findings were within normal limits.

The patient was not premedicated. She was monitored with a pulse oximeter, nasopharyngeal temperature probe, electrocardiogram, and an automated non-invasive blood pressure machine. A peripheral iv cannula (24G) was inserted and iv infusion of 0.2% NaCl in 5% dextrose solution was initiated at 8 ml.kg-1.h-1. The induction of anaesthesia was performed with 25 mg fentanyl and 30 mg propofol, and after checking the lungs were ventilated through a mask without difficulty. neuromuscular monitoring was performed by Datex Relaxograph and 10 mg atracurium iv was administered. Intermittent positive pressure ventilation of the lungs was maintained with 100% oxygen through a face mask. As the TOF ratio reached zero, tracheal intubation was easily performed at the first attempt. Anaesthesia was maintained with propofol infusion (10 mg.kg-1.h-1 during the first 10 minutes, and then 8 mg. kg-1.h-1 during the second 10 minutes, and thereafter 6 mg. kg-1. h-1) by an infusion pump (IVAC 770, USA). She was monitored using a capnograph and mechanically ventilated with 50% O2 and air to maintain end tidal CO2 at 35 mmHg. 10 mg fentanyl 30 minutes after the induction and three additional intermittent bolus doses of atracurium (3 mg each) when T1 recovered to 25% of the control value, were administered. The operation ended approximately 95 minutes after the induction of anaesthesia and propofol infusion was stopped approximately 10 minutes before the end of surgery. At the end of the operation 0.5 mg neostigmine and 0.25 mg atropine iv were used to reverse the residual neuromuscular block. Tracheal extubation and recovery were uneventful.

DISCUSSION

There is little clinical information in the literature about the anaesthetic management of the patients with Cornelia de Lange syndrome. A high proportion of these patients are mentally retarded and may show behavioura disturbances (1,2). Mentally retarded patients may be difficult to manage during the induction of anaesthesia and they may benefit from sedative premedication. However, these patients need close follow-up after a sedative premedication because responses to drugs may be unpredictable due to immaturity of organ systems (3).

Reported anaesthetic experience with these patients highlights the potential for aspiration and difficulty with tracheal intubation (3-5). In the patients who possibly have difficulty with tracheal intubation, regional anaesthesia would be the preferred option. Lumb and Carli (6) reported a case with Cornelia de Lange syndrome who had respiratory arrest after a caudal injection of bupivacaine. However, it most likely resulted from an accidental injection of bupivacaine in the subarachnoid space, so regional anaesthesia should not be implicated in that case (7,8).

If general anaesthesia is a necessity as in the present case, assessment of the ability to assist ventilation by a mask without depressing spontaneous ventilation, irrespective of whether anaesthesia is induced by iv or inhalation method, may be a safe approach. Insertion of a peripheral intravenous cannula is not always easy if the patient is a child, and there may be additional difficulty when the child is mentally retarded. However an intravenous line is essential for the safety of the patients in emergencies like seizure or a respiratory arrest, which were highlighted in the literature as being associated with Cornelia de Lange syndrome (3-5). An intravenous line can be inserted more easily when these children are sedated properly or accompanied by a parent who can calm them down.

After a peripheral intravenous cannula is inserted, anaesthesia can also be induced with intravenous anaesthetics. Intravenous induction of anaesthesia is essential for the anaesthetic management of a patient who has musculoskeletal disorders. Patients who are susceptible to develop malignant hyperthermia usually present with musculoskeletal disorders (9). In paediatric anaesthesia, propofol provides satisfactory and stable anaesthesia with rapid and complete recovery, and less nausea and vomiting even after repeated doses or continuous infusion (10). Propofol is also a safe anaesthetic agent in malignant hyperthermia susceptible patients (11). In the present case propofol provided a smooth induction and maintained a steady anaesthesia.

We preferred a nondepolarizing muscle relaxant, atracurium, to facilitate endotracheal intubation and to maintain muscle relaxation. Succinylcholine should not be a muscle relaxant used in paediatric anaesthesia unless it is essential to secure the airway rapidly. There are many reports about the adverse cardiovascular reactions of succinylcholine in paediatric anaesthesia (12,13) and it also has been implicated in triggering malignant hyperthermia (9). Therefore, a nondepolorizing muscle relaxant seems to be the best choice in paediatric anaesthesia, especially if the patient has musculoskeletal disorders. Sargent (3) used vecuronium uneventfully in a patient with Cornelia de Lange syndrome and we used atracurium in the present case without any problem.

In conclusion, mental retardation, difficult endotracheal intubation, and malignant hyperthermia are the key factors to be considered in anaesthetic management of a patient with Cornelia de Lange syndrome. Total intravenous anaesthesia with propofol, fentanyl and atracurium seems to be a good choice in these patients when general anaesthesia is a necessity.

Correspondence to : Ertan ÖZTÜRK, M.D.
3. Cad. 36/5
Bahçelievler
06500 ANKARA - TÜRKİYE
Phone : 0 312 - 221 04 10

REFERENCES

1. Joubin J, Pettrone CF, Pettrone FA. Cornelia de Lange's syndrome. A review article (with emphasis on orthopedic significance). Clin Orthop 1982; 171: 180-185.
2. Filippi G. The de Lange syndrome: Report of 15 cases. Clin Genet 1989; 35: 343-3463.
3. Sargent WW. Anesthetic management of a patient with Cornelia de Lange syndrome (Letter to the Editor). Anesthesiology 1991; 74: 1162-1163.
4. Veall GRQ. An unusual complication of Cornelia de Lange syndrome. Anaesthesia 1994; 49: 409-410.
5. Kiriyama M, Masuda A, Satone T, Higbuchi A, Ito Y. The anesthetic management of a patient with Cornelia de Lange syndrome. Masui 1984; 33: 1392-1394.
6. Lumb AB, Carli F. Respiratory arrest after a caudal injection of bupivacaine. Anaesthesia 1989; 44: 324-325.
7. Fortuna A. Respiratory arrest after a caudal injection of bupivacaine (Letter to the Editor). Anaesthesia 1989; 44: 1007.
8. Lee E, Collins G. Respiratory arrest after caudal bupivacaine (Letter to the Editor). Anaesthesia 1990; 45: 63-64.
9. Strazis KP, Fox AW. Malignant hyperthermia: A review of pubished cases. Anesth Analg 1993; 77: 297-304.
10. Hannallah RS, Britton JT, Schafer PG, Patel RI, Norden JM. Propofol anaesthesia in paediatric ambulatory patients: a comparison with thiopentone and halothane. Can J Anaesth 1994; 41: 12-18.
11. McKenzie AJ, Couchman KG, Pollock N. Propofol is a 'safe' anaesthetic agent in malignant hyperthermia susceptible patients. Anaesth Intensive Care 1992; 20: 165-168.
12. Delphin E, Jackson D, Rothstein P. Use of succinylcholine during elective pediatric anesthesia should be reevaluated. Anesth Analg 1987; 66: 1190-1192.
13. Rosenberg H, Gronert GA. Intractable cardiac arrest in children given succinylcholine (Letter to the Editor). Anesthesiology 1992; 77: 1054.