Malignant Hyperthermia in a Three-Year-Old Child with Microstomia

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INTRODUCTION

Malignant hyperthermia (MH) is an autosomal dominant pharmacogenetic disorder that presents as a hypertensive response to volatile, halogenated anesthetics and succinylcholine. MH has been directly associated with two diseases: central core disease and King-Denborough syndrome. 1 Associations with other disorders have been suggested, but direct linkage to MH has been difficult to prove. One of those disorders is Freeman-Sheldon syndrome (multiple congenital contracture syndrome), 2,3 Clinical features of Freeman-Sheldon syndrome (FSS) include flexion contracture of the fingers with abduction of the thumbs, kyphoscoliosis, talipes equinovarus, and developmental delay. Characteristic facial features include a flat midface, long philtrum, microstomia, and a whistling shape to the lips. We present a case of malignant hyperthermia in a three-year-old girl with microstomia but no other features of Freeman-Sheldon syndrome.

CASE REPORT

The patient was a three-year-old, healthy, active girl with microstomia that presented for elective cleft lip repair. The physical examination was normal with the exception of a very small mouth opening. The mother was also told that the child has a narrow or restrictive right nasal passage. The patient had never had previous surgery or anesthesia. There was no history of MH in the immediate or extended family.

Oral midazolam (0.5mg/kg) was administered 25 minutes prior to induction of anesthesia. Anesthesia was induced with facemask with sevoflurane in oxygen. A 22-gauge intravenous catheter was inserted in the left hand. After an adequate depth of anesthesia was obtained with the patient breathing spontaneously, a fiberoptic-assisted nasotracheal intubation was performed without difficulty with a 4.0mm ID nasal RAE tracheal tube. The end-tidal CO2 after tracheal intubation was greater than 80 mmHg. The nasopharyngeal temperature was 37.1°C. As the surgeon needed access to the nose, the temperature was moved to the right axilla (36.2°C). Despite vigorous hyperventilation, the end-tidal CO2 remained at 75 mmHg. At this time, it was felt that an arterial blood gas was indicated. The surgical drape over the right leg was lifted and marked contracture of the right leg was noted. The surgeon was alerted that an acute MH episode was occurring. He immediately began closing the incision. In rapid succession, dantrolene (3 mg/kg iv) was administered, an arterial catheter was inserted, central line fentanyl was inserted into both limbs of the anesthesia circuit, and surface cooling was begun. The initial blood gas revealed a mixed respiratory and metabolic acidosis with a pH of 7.037 and a base deficit of -14. Three ml/kg of NaHCO3 was administered intravenously. Soon after the administration of dantrolene, the end-tidal CO2 decreased from 75 to 55 and then 37 mmHg. After stabilization in the operating room, she was transported to the recovery room intubated and maintained on assisted ventilation. Sedation was accomplished with intravenous propofol 75 mg/kg/min. Her blood pressure, heart rate, and temperature were normal during the next three hours and serial arterial blood gases were normal. She was weaned from assisted ventilation and extubated without difficulty.

DISCUSSION

Malignant hyperthermia is a rare but potentially fatal disease especially in patients with no previous history of MH. The gold standard for diagnosis of MH susceptibility is exposure to a fresh muscle biopsy specimen to different concentrations of halothane and caffeine: the halothane-caffeine contracture test. This process is invasive and expensive. Despite many years of research, no less invasive test has been developed. Preoperative knowledge of disorders associated with an increased likelihood of MH can guide preparation for surgery and increase vigilance for early signs of MH.

There are a number of diseases that may be associated with MH and other diseases may have a coincidental association with MH. Differentiating true predisposition from coincidental association is not easy.

Freeman-Sheldon syndrome is a rare form of multiple congenital contracture syndrome described by Freeman and Sheldon in 1938. It is also referred to as distal arthrogryposis type 2A, craniorumpotarsal dysplasia or whistling face-windmill vane hand syndrome. 4 The syndrome is limited to the musculoskeletal systems with primary deformity from myopathic arthropathy. The strict diagnostic criteria for FSS include two or more major defects plus microstomia, whistling face, prominent nasolabial creases and ‘‘M-shaped’’ chin dimple. Major defects include ulnar deviation of wrists and fingers, camptodactyly, hypoplastic and/or absent flexion creases, and/or overriding fingers at birth, talipes equinovarus, calcaneovalgus, vertical talus, and/or metatarsus varus. Patients with FSS constitute several anesthetic challenges, such as the difficult airway, poor venous access, perioperative pulmonary complications, and susceptibility to MH. 5 6 7 8 9 10 The underlying myopathy of FSS may predispose patients to MH. There are two case reports of patients with FSS who developed MH after induction of anesthesia using halothane and succinylcholine. 4

Thus far, there are no case reports on isolated microstomia associated with MH. Our three-year-old patient has isolated microstomia without association with Freeman-Sheldon syndrome. After induction with sevoflurane and oxygen, she developed malignant hyperthermia, which was responsive to a single dose of dantrolene. After the MH episode, there was no ongoing myopathy with normal CK level, indicating that she has no underlying myopathy. In addition, genetic testing was negative for FSS. This disorder of isolated microstomia may have a coincidental association with MH. However, it is difficult to distinguish this coincidental association of isolated microstomia from true predisposition like that of Freeman-Sheldon syndrome. More cases are needed to be reported for clarification.

REFERENCES