ANESTHESIA FOR A PATIENT WITH OSTEOGENESIS IMPERFECTA, ACHONDROPLASTIC DWARFISM AND HISTORY OF MALIGNANT HYPERTHERMIA

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Abstract

A primary goal for anesthesia providers is to maintain patient safety. This is an even greater concern when taking care of a patient with a complicated medical history. This case report, discusses the care of a 47 year-old female patient who presented to a tertiary care center for an orthopedic procedure. Her medical history included osteogenesis imperfecta (OI), achondroplastic dwarfism and suspicion of malignant hyperthermia (MH). There were multiple anesthetic implications to ensure safety for this patient during the perioperative period. OI concerns include bone fragility and potential for multiple fractures even after inoffensive trauma. Achondroplastic dwarfism concerns include abnormalities of the upper airway and difficulty with visualizing the glottic opening during direct laryngoscopy.1 Malignant Hyperthermia is a life threatening disorder, which places the patient at risk for a hypermetabolic reaction if exposed to select anesthetic agents.

Introduction:

Patients with osteogenesis imperfect (OI) are placed at high risk during anesthesia for both physiological and anatomical reasons. Complications include osteoporosis, joint laxity, and tendon weakness.[2] Pulmonary compromise may also occur if the patient displays thoracic distortion. These patients have an elevated basal metabolic rate that causes an increase in core body temperature and can mistaken to be MH. No consistent evidence has shown that OI is always associated with MH.[2] Achondroplastic

dwarfism is the most common form of dwarfism occurring at the rate of 1:30,000 live births. Airway abnormalities such as macroglossia, micrognathia, small oral opening and temporomandibular joint immobility can make mask ventilation and intubation challenging for anesthesia providers. The perioperative period can also be complicated with managing restrictive lung disease patterns and cardiovascular abnormalities.[1] Malignant hyperthermia (MH) is a hypermetabolic disorder that is triggered by anesthetic agents such as succinylcholine, or inhaled anesthetics: halothane, isoflurane, sevoflurane and desflurane. Several musculoskeletal disorders are coupled with MH including the patient's diagnosis of osteogenesis imperfecta. Identifying susceptible patients prove difficult because it is a silent disorder until triggered. In addition, 50% of patients who experience an MH crisis had previously received a triggering agent without showing symptoms.[3]

In this case report a patient presents to a tertiary care hospital with all three of these conditions. Naturally, the anesthetic plan for this patient sparked a large amount of discussion for anesthetic management and concern for patient safety.

Case Report

A 47 year-old Caucasian female presented to a tertiary care center for her 55th surgery. She was 36 inches tall and weighed 24 kg for a BMI of 28.2. The patient was an achondroplastic dwarf with a past medical history of OI leading to her many hospital admissions and surgical history. The patient was wheelchair bound and presented with an adduction deformity of the right hip. The procedural goal was palliative to remove the constant pressure points that caused pain between both knees. The patient also had a suspected history of MH due to parental report of "anesthetic complications" from a previous procedure. Medications included cephalexin, alprazolam, and oxycodone. She had no known allergies.

The patient was seen in the preoperative holding area by the

anesthesia team for the preoperative assessment. The assessment focused on her airway, cardiopulmonary and musculoskeletal status. Key airway assessment findings included a Mallampati class I, oral opening was greater than 4 cm, full dentition intact, mandibular length adequate, thyromental distance of 6 cm, cricothyroid membrane palpable and an atlanto-occipital extension of less than 35 degrees. The patient's head shape and size was normal and her neck thin. The patient experienced constant pain in her left leg from pressure point contact. She preferred the left lateral position with a pillow between her knees to prevent pressure. Her chest was smaller than expected for an adult female and had a kyphoscoliotic shape. Lung sounds clear to auscultation and chest x-ray clear throughout lung fields. Heart rate assessed as regular with normal S1 and S2 pattern. Electrocardiography revealed sinus rhythm with short PR intervals.

With the inconclusive malignant hyperthermia history, the anesthesia team proceeded to follow the MH protocol. All MH triggers were removed from the OR with a total intravenous anesthetic planned. Risks and benefits of the procedure and anesthetic technique were discussed with the patient. She accepted the discussion and signed, giving informed consent. The patient's number of previous surgical procedures places her at risk for blood loss. Blood consent was obtained with two units of packed red blood cells available. Preoperative intravenous access was obtained with a 20 gauge IV placed in the right forearm. The patient is a Patient Status 3.

The anesthesia workstation was prepared in accordance with Malignant Hyperthermia Association of the United States (MHAUS) recommendations. All MH triggers needed to be eliminated including removing succinylcholine syringes from the induction drug set up and disabling the vaporizers by taping them to the "off" position. The carbon dioxide (CO_2) absorbent was changed to prevent any fractionated inspired agent from

entering the breathing circuit. The anesthesia gas machine (AGM) was flushed with 10L/min of oxygen for 20 minutes[4] and a new breathing circuit was installed on the AGM. Please see Figure 1.

The patient was taken to the OR and given 1 milligram of midazolam and 25 micrograms of fentanyl for preoperative sedation and pain management. The patient was lifted to the OR table to prevent discomfort and then secured to the table with safety straps. Initial position was supine with blankets to support her spine. Care was taken to support her arms on padded arm boards bilaterally at less than a 90-degree angle. Standard monitors were placed and vital signs (VS) recorded. The patient was denitrogenated with 100% oxygen and induction of anesthesia began with 50 micrograms of fentanyl, 60 milligrams of lidocaine, and 50 milligrams of propofol. Once unconscious ventilation was confirmed, the patient received 20 milligrams of rocuronium bromide for paralysis and was intubated with a Macintosh 3 blade after Cormack I direct visualization of the cords. The patient was intubated with a pediatric cuffed 5.0 endotracheal tube. A propofol infusion at 120 microgram/kg/min was initiated and a right internal jugular triple lumen catheter was placed. A Hotline™ (Dublin, OH) fluid warming system was connected to the new central line for fluid management and warming. An upper body forced air warming blanket was applied Bair Hugger® Therapy from Arizant Inc. The patient was given incremental fentanyl boluses (1 microgram/kilogram) throughout the procedure.

As requested by the surgeon, further muscle relaxation was withheld in order to monitor motor responses during the procedure. Patient was given Zofran 4 milligrams, given to prevent the female predisposition to postoperative nausea and vomiting with general anesthesia 30 minutes prior to emergence. The patient regained four train of four twitches and was reversed with 2 milligrams of neostigmine and 0.4 milligrams of glycopyrrolate. Upon emergence the patient awoke, opened her eyes, followed

commands, and was breathing spontaneously. However, she displayed less than 5 milliliters/kilogram tidal volumes and was transferred to the post anesthesia care unit with an endotracheal tube and on oxygen through a t-piece to protect her airway. Postoperative vital signs included a temperature of 37.3 degrees Celsius(C), 130 heart rate, blood pressure 120/80, regular respiratory rate of 16 and saturations of 100% on 40% oxygen. She gradually gained strength and was extubated.

Discussion

The pathophysiology of osteogenesis imperfecta is decreased collagen synthesis. Ninety percent of individuals have mutations on the type I collagen genes, pro-a1 or pro- a2. A phenotype often indistinguishable from OI type II or III is a mutation of two of the genes responsible for encoding proteins of an enzyme complex LEPRE1 and cartilage-associated protein causing an autosomal recessive OI.[1] Patients with osteogenesis imperfect are placed at high risk during anesthesia for both physiological and anatomical concerns. Physical manifestations are a large head, small bowed limbs, short neck, blue sclera, otosclerosis, and brittle teeth. Complications lend themselves to osteoporosis, joint laxity, and tendon weakness.[2] These patients may also have abnormal platelet function leading to an increased risk of bleeding. There is a tendency for early development of atherosclerosis so a cardiovascular assessment that includes evaluation of the mitral and aortic valve function is important. Pulmonary compromise may also occur if the patient displays thoracic distortion. These patients have an elevated basal metabolic rate that causes an increase in core body temperature and can mistakenly be thought to be MH. There has been no consistent evidence that OI is always associated with MH.[2]

Achondroplastic dwarfism is a phenotype of disproportionate stature. In 95%of patients the same point mutation occurs on the gene that encodes for fibroblast growth factor receptors, FGFR3.[1] The anesthetic management of these patients proves a

challenge with abnormalities of their upper airways and difficulty visualizing the glottic opening on direct laryngoscopy. Intubation is often challenged by characteristics of a short neck, protruding tongue, as well as enlarged tonsils and adenoids.[1] Further, these patients may have subglottic stenosis, tracheal and bronchial narrowing. A thoracic dystrophy may be present causing the patient to have reduced lung volumes due to the restrictive disease pattern. Cardiac dysfunction may include acquired valvular disease, cor pulmonale and cardiomyopathy. It is advisable to preoperatively obtain a chest x-ray, echocardiogram as well as a physical exam to evaluate the presence and severity of these conditions.[1] For the possibility of pulmonary and cardiac complications it is imperative to avoid hypoxemia and hypovolemia. This can exacerbate existing pulmonary hypertension and worsen right ventricular function.[1]

Malignant hyperthermia is a hypermetabolic disorder that is triggered by anesthetic agents such as succinylcholine, or inhaled anesthetics: halothane, isoflurane, sevoflurane and desflurane. Several musculoskeletal disorders are associated with MH including osteogenesis imperfecta. Exposure to a triggering agent can cause a dramatic increase in skeletal muscle metabolism. The most common first sign of MH is a rapid increase in end tidal carbon dioxide ($C0_2$) as metabolism accelerates. Other early signs are tachycardia, tachypnea, and increased oxygen consumption, acidosis, muscle rigidity, and rhabdomylosis.[5] Further symptoms are unstable blood pressure, cyanosis, mottled skin, diaphoresis, dysrhythmias and an increase in patient's body temperature. Temperature increase can be as much as 1-2 degrees C every 5 minutes. The mortality rate of unrecognized MH can be as high as 80%.[6] Identifying susceptible patients is difficult because it is a silent disorder until triggered. In addition 50% of patients who experience an MH crisis had previously received a triggering agent without showing symptoms.[3] MH is a hypermetabolic syndrome that occurs from an abnormal amount of calcium release by the sarcoplasmic reticulum in to the sarcoplasm.

Increased levels of calcium cause elevated oxygen consumption and anaerobic metabolism. The transportation of the calcium is mediated by the ryanodine receptor, isoform 1 (RYR1).[3] About 50% of known MH cases are caused by mutations on chromosome 19 at the RYR1 receptor. A small percentage is dihydropyridine (DHP) receptor on chromosome 1.[3] The most current dependable method to confirm diagnosis is a muscle biopsy test where the muscle fibers are placed in contact with caffeine and halothane.[7] There is 95% reliability for patient susceptibility with a positive muscle contraction.[6]

Summary

This case report presents a patient with three rare disorders that each has multiple anesthetic implications. The diagnosis of osteogenesis imperfecta predisposes the patient to brittle bones, joint laxity, and tendon weakness. Safety in positioning, movement and transfer of the patient becomes extremely important for a patient with OI. Furthermore, these patients often have altered pulmonary or cardiovascular function. Achondroplastic dwarfism has additional implications that include airway abnormalities that make mask ventilation and direct laryngoscopy challenging. The presence of the difficult airway cart and availability of immediate surgical personnel are two prudent measures to be taken prior to induction. Additionally, this patient reported a possible MH exacerbation during an earlier surgical procedure. This mandates preparation of the anesthesia workstation and clarification amongst anesthesia personnel in order to ensure that no trigger agents reach the patient and that the team is prepared in the event of a hypermetabolic event. This case illustrates the importance of a thorough preoperative assessment, development of a comprehensive plan involving both anesthesia and surgical teams and the need for constant vigilance. Maintaining an open line of communication throughout the procedure is imperative in order to reach the ultimate goal of patient safety.

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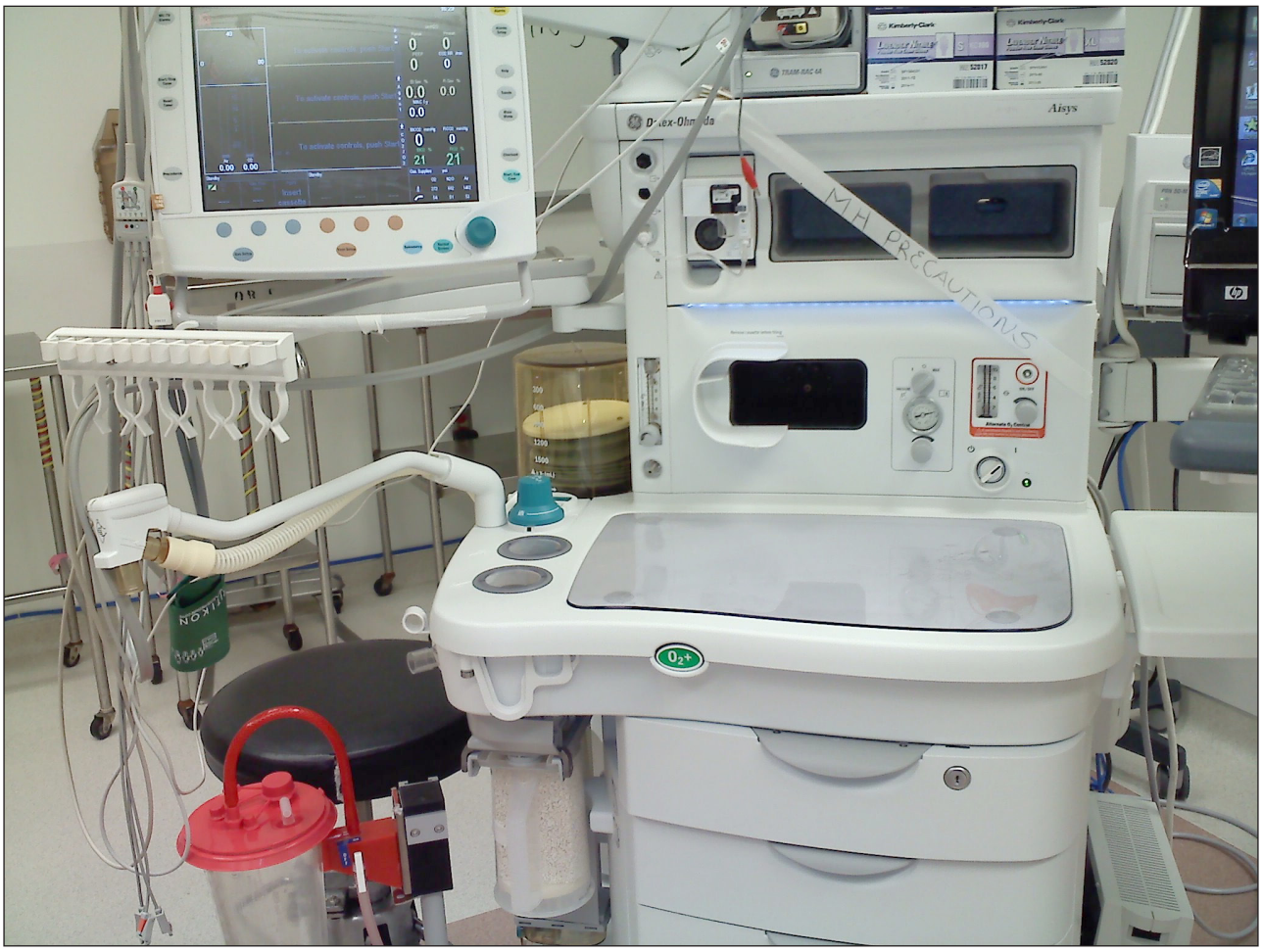
Acknowledgements

Acknowledgment is given to John M. O'Donnell DrPH, RN, CRNA, MSN as mentor and critiquing the case study.

FIGURE 1



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