Anesthetic Management Of A Patient With Tetra-Amelia Syndrome

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Anesthetic Management Of A Patient With Tetra-Amelia Syndrome

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Abstract

Tetra-amelia Syndrome is a very rare autosomal genetic disorder characterized by the complete absence of all extremities with associated anomalies. Anesthetic management of these patients may be challenging, including problems with vascular access, lack of sites available to monitor blood pressure and possible difficult airway. We describe the anesthetic management of a 5 month-old with Tetra-amelia Syndrome. The problems and options in obtaining intravenous access, securing the airway and monitoring vital signs during the procedure are discussed. The use of near infrared spectroscopy for hemodynamic monitoring is described to estimate adequate organ perfusion when blood pressure measurement is not practical.

Introduction

Tetra-amelia Syndrome (TS) is a very rare autosomal genetic disorder with heterogeneity in some patients characterized by the complete absence of all extremities (“tetra” meaning “four”, and “amelia” meaning the failure of an extremity to develop before birth) with associated craniofacial, cardiopulmonary, ocular, neurological and urogenital anomalies (Table 1) [1-3]. Although there are two case reports of patients with tetra-amelia who underwent surgical procedures under anesthesia, none has been reported for TS [4, 5].

In this case report of patient with TS the anesthetic challenges related to the intravenous access, drawing of blood for laboratory tests, airway management and monitoring of vital signs are discussed. In this patient near infrared spectroscopy (NIRS) was used for hemodynamic monitoring. The trends of cerebral regional oxygenation saturation were used to ensure adequate cerebral oxygenation and as an alternative to blood pressure measurements to estimate adequate organ perfusion.

Case Report

A 5 month-old, 3.6 kg male infant was scheduled for Nissen fundoplication and gastrostomy tube placement for reflux and poor feeding. The patient was born at 38 weeks gestational age and weighed 1.8 kg. His past medical history was significant for TS, including dextroscoliosis, syrinx of the spinal cord, malrotated kidneys, and a patent foramen ovale. The infant had also had chronic recurrent pneumonias. He had been hospitalized previously for respiratory distress and dehydration. Cytogenetic analysis did not reveal any evidence of a consistent detectable numerical or structural chromosomal abnormality. Our preoperative examination revealed an infant with a large occiput, smooth facies, long philtrum, micrognathia, a beak-shaped nose and complete absence of all extremities.

Once in the operating room, electrocardiogram leads, precordial stethoscope and skin temperature probes were placed. The pulse oximetry probe was placed on the patient’s earlobe. Neither invasive nor noninvasive blood pressure monitoring was possible because of complete absence of all extremities. The femoral arteries were not palpable. Since significant blood loss and volume shifts were unlikely during the procedure, we used NIRS to follow regional cerebral and somatic oxygen saturation (rSO2) trends as means of estimating adequate cerebral and somatic oxygenation [6]. INVOS 5100 C (Pediatric Somanetics Corporation, Troy, MI) probes were placed on the fronto-temporal area (cerebral) and posterior lumbar area (renal). Baseline regional oxygenation saturation (rSO2) was measured when the patient was awake before induction. Inhalation induction was achieved with 30% oxygen, 70% nitrous oxide and sevoflurane. For intravenous access, we were able to place a 24-guage catheter in a scalp vein. The patient received 10 mg of intravenous propofol and 4 mg of rocuronium to achieve adequate relaxation for endotracheal intubation. The airway was secured with a 3.0 cuffed endotracheal tube and anesthesia was maintained with sevoflurane and 15 mcg of intravenous fentanyl. The patient was mechanically ventilated. The infant’s ECG, oxygen saturation, end tidal CO2 and NIRS stayed within the normal range except for one episode in which rSO2 dropped from his baseline of 89% to 85%; this was treated with a fluid bolus. The case finished uneventfully with minimal blood loss. The neuromuscular block was reversed. The patient was extubated and sent to the post anesthesia care unit.
(PACU) in stable condition. NIRS monitoring was continued in the PACU and was discontinued before the patient went to the floor.

Discussion

Tetra-amelia is extremely rare. It may exist alone or with multi-organ involvement as in Tetra-amelia Syndrome (Table 1). It occurs as a result of a mutation in the WNT3 gene located on the 17q21 chromosome loci [2]. Mutation of this gene inhibits the production of functional WNT3 protein causing disruption in the normal formation of extremities and other birth defects [2, 7]. The parents of a child with TS are heterozygotes (each carries one copy of the mutated gene) but they do not develop the syndrome. Our patient did not show any chromosomal abnormalities, which is similar what was reported by Sousa, et al. and Krahns, et al. [1, 8]. Sousa, et al. reported “tetra-melia and lung hypo/aplasia syndrome” as a distinct autosomal recessive condition with no identified gene [1]. Also, Krahns, et al. suggested genetic heterogeneity for TS [8]. TS has been reported in people of different ethnic groups and there have been reports of parental consanguinity in some cases [1, 2, 8].

TS may be confused with thalidomide embryopathy (limb abnormalities, facial abnormalities cardiac malformations, poor fetal growth, urogenital, gastrointestinal and spinal defects), Roberts Syndrome (symmetric limb defects, growth retardation, mental retardation, and craniofacial abnormalities) and Odontotrichomelic Syndrome (severe absent malformations of all extremities, abnormal teeth, malformation of ears, mental, thyroid, electroencephalographic and ECG abnormalities) [9 -11]. TS can be detected prenatally during ultrasonography and aid in the assessment of other malformations. Cytogenetic testing of DNA obtained with amniocentesis can confirm the diagnosis. The clinical course of the disease, management, complications and prognosis is quite variable and seldom described in the literature as the disease is so rare.

Until now there has been no published case report of a patient with TS who underwent surgery and anesthesia, but there are case reports of two adult patients with tetra-amelia-like defects who underwent surgery and anesthesia [4, 5]. In the first case, the patient did not have complete tetra-amelia, as she had a partial right upper extremity with enough area to place a blood pressure cuff [4]. The second case performed under spinal anesthesia was not TS but rather acquired tetra-amelia as the patient lost his extremities because of Buerger’s Disease. For this patient, noninvasive blood pressure monitoring was not possible and femoral arteries were not palpable. The authors (Ohtsuka et al.) opted to palpate and check the carotid artery intermittently and monitor consciousness during the entire procedure [5]. This was similar to our patient, since only the carotid arteries were palpable and there was no extremity available for a blood pressure cuff; however, in our case regional anesthesia was not practical or feasible both because of the nature of the surgery and the age of our patient.

The major issues in perioperative management of a patient with TS include possible difficult airway, intravenous access and monitoring of blood pressure. These patients may have a high and narrow palate, macrostomia, and micrognathia besides other craniofacial abnormalities that make airway management challenging. Peripheral venous intravenous access may be obtained from non-extremity sites such as the scalp and neck. Central venous access from the internal jugular, is an alternative option [4]. Central venous access from subclavian or femoral veins has not been reported in these patients as these vessels may be absent or poorly developed. However ultrasound can be used to assess the femoral vessels if they are suitable for central catheter placement. Pulse oximetry probes are placed on the ears, lips, tongue, nose or forehead in these patients [3]. For forehead pulse oximetry, reflection (backscatter) sensors are used since transillumination techniques cannot be employed [12, 13].

Blood pressure is an important monitoring modality for accessing adequate cerebral perfusion in a patient under anesthesia. The brain, being a high oxygen extraction site, is most vulnerable to ischemia when its blood supply is compromised. Since non-invasive blood pressure monitoring cannot be done in patients with TS for lack of a site to place a blood pressure cuff, another option would be an invasive intra-arterial blood pressure monitoring [4]. No case reports of intra-arterial blood pressure monitoring from femoral or axillary arteries has been reported, presumably because these vessels are not developed. It can be a dilemma for the anesthesiologist to perform this invasive intra-arterial cannulation for a short diagnostic or surgical procedure with minimal blood loss and minimal fluid shifts considering the risks associated with such procedure. However, anesthesia providers are left with no other alternatives to measure the blood pressure perioperatively. Although it cannot replace blood pressure monitoring,
NIRS is an option in cases such as ours. NIRS is a non-invasive, optical technique used to estimate tissue oxygenation. It provides real-time estimates of regional oxygen saturation (rSO2) in the cerebral and somatic tissue beds. Multi-site monitoring is done to compare the oxygen extraction. Renal rSO2 is typically 15-20% higher than brain rSO2, as the brain is a high oxygen extraction organ while the kidney has low oxygen extraction. [14]. A decrease in rSO2 might result from reduced arterial blood flow, reduced oxygen saturation in the arterial blood, increased oxygen consumption, hypotension, blood loss, decrease in PCO2 and venous congestion [15,16]. Hoffman, et al. found that an rSO2 value of less than 40-50% or a change in baseline greater than 20% is associated with hypoxic–ischemic neural injury [14]. A sustained drop in rSO2 of cerebral NIRS is related to ischemic events that could be due to hypotension or blood loss [15].

There is good correlation between NIRS parameters and blood loss. Torella, et al. concluded that NIRS is potentially useful technique for monitoring blood loss in humans [16]. Although NIRS cannot be a substitute for blood pressure monitoring, it is a useful tool to monitor adequate organ perfusion when there are no other available practical and reasonable alternatives. Transthoracic or esophageal echocardiography may be another alternative especially in cases in which massive blood loss and significant hemodynamic changes are expected. Intraoperative echocardiography provides important information including immediate assessment of volume status, cardiac output, cardiac contractility and myocardial ischemia [17].

Conclusion

The anesthetic management of a patient with TS can be challenging. Problems related to vascular access, lack of sites available to monitor blood pressure by invasive or non-invasive techniques and possible difficult airway may be encountered. NIRS is a non-invasive monitor that can provide anesthesiologists important information to help manage a patient’s hemodynamic status intraoperatively.

References


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