Perioperative anaesthetic management of patient with amelia and phocomelia - case report

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Abstract:

Phocomelia is a congenital condition involving malformations of the arm and leg, resulting in a fin-like appendage. It is often caused by the mother's use of the drug thalidomide during pregnancy, with various mechanisms being proposed. There is no specific treatment for phocomelia or amelia, but in certain cases surgical intervention may be recommended to improve limb function or other functional outcomes. This case report concerns a ten-year-old female patient with congenital amelia of the upper extremities and phocomelia of the lower extremities who was admitted to the hospital for elective thoracic scoliosis surgery It is a demanding and long-term operation with estimated major blood loss. In addition, the prone position of the patient makes access difficult in case of resuscitation, and therefore the role of the anesthesiologist in the care of such patients is crucial in the perioperative period. When caring for such patients, there are many technical difficulties, starting with the monitoring of arterial pressure values. Furthermore, difficulties may arise related to peripheral venous access, which can be a great challenge, and the placement of a central venous catheter is sometimes indicated. Also, intubation can be difficult due to limited mobility of the cervical spine. Early and adequate preoperative assessment of the patient is essential for the induction and maintenance of safe anesthesia and postoperative care. The current review of the literature does not reveal strict recommendations on the optimal treatment of these patients. Due to all the above-mentioned difficulties, preoperative preparation and assessment are of key importance for the introduction and maintenance of safe anesthesia, as well as postoperative care.

Keywords: phocomelia, amelia, scoliosis, anaesthetic management, anaesthesia

Sažetak:

Perioperativna anesteziološka skrb pacijenta sa amelijom i fokomelijom - prikaz slučaja

Fokomelija je kongenitalno stanje koje uključuje malformacije ruku i nogu, što rezultira privjeskom poput peraje. Često je uzrokovana majčinom uporabom lijeka talidomida tijekom trudnoće, uz predložene različite mehanizme objašnjenja. Ne postoji specifično liječenje fokomelije ili amelije, ali u određenim slučajevima može se preporučiti kirurška intervencija za poboljšanje funkcije ekstremiteta ili drugih funkcionalnih ishoda. U ovom prikazu slučaja radi se o desetogodisnjoj bolesnici sa kongenitalnom amelijom gornjih ekstremiteta i fokomelijom donjih ekstremiteta koja je primljena je u bolnicu na elektivnu operaciju torakalne skolioze. Radi se o zahtjevnom i dugotrajnom operativnom zahvatu sa velikim procijenjenim gubicima krvi. Osim toga, potrbušni položaj bolesnika otežava pristup u slučaju reanimacije te je stoga uloga anesteziologa u zbrinjavanju ovakvih bolesnika ključna u perioperativnom razdoblju. Kod zbrinjavanja ovakvih bolesnika postoji mnogo tehničkih poteškoća počevši od monitoriranja vrijednosti arterijskog tlaka, a mogu se pojaviti poteškoće vezane uz periferni venski pristup

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što može predstavljati veliki izazov te je ponekad indicirano postavljanje centralnog venskog katetera. Također, intubacija može biti otežana zbog ograničene mobilnosti vratne kralježnice. Rana i adekvatna prijeoperacijska procjena bolesnika ključna je za indukciju i održavanje sigurne anestezije i postoperativne skrbi. Trenutačno pregledom literature ne nalaze se stroge preporuke o optimalnom liječenju ovih bolesnika. Zbog svih navedenih poteškoća, prijeoperacijska priprema i procjena od ključne su važnosti za uvođenje i održavanje sigurne anestezije, kao i postoperativne skrbi.

Ključne riječi: fokomelija, amelija, skolioza, anesteziološka skrb, anestezija

Introduction

Phocomelia is a congenital condition that involves malformations of human arms and legs which results in a flipper-like appendage, while amelia is congenital absence of arms or legs or both (1). A prominent cause of phocomelia is the mother's use of the drug thalidomide (which was marketed for treating anxiety and morning sickness) during pregnancy; however, the causes of most cases are to be determined (2). Various explanatory mechanisms have been proposed, including genetic mutations, teratogenic exposures during embryonic development as mentioned above and environmental influences (3). Aside from the teratogenicity of thalidomide, researchers have assumed that phocomelia may be caused by anomalous origins of the subclavian artery (4). There is no specific treatment for phocomelia nor amelia, but in certain cases, a surgical intervention may be recommended for improving limb function or other functional outcomes (5). The anesthetist faces a variety of difficulties, particularly when treating patients with phocomelia. Some of these challenges include difficulty in monitoring blood pressure, venous access, difficult intubation and fluid management. Also spinal abnormalities, such as spina bifida, scoliosis, and anterior fusion of the thoracic and lumbar vertebrae, may make spinal or epidural anesthesia more difficult (6).

Case report

A 10 year old patient with congenital amelia of upper extremities and phocomelia of lower extremities was admitted to the hospital for elective surgery of thoracic scoliosis. Besides, the patient had suffered from vesicoureteral reflux of 2nd grade as well as from urovaginal reflux. Phocomelia patients have mobility restrictions and require assistance in transport to theatre. Scoliosis surgery is a demanding and long-term operation with estimated major blood loss. A study conducted by Xuerong Yu and associates has shown that more than half of the patients (59.7%) undergoing scoliosis surgery had massive blood loss (7). In addition, the prone position of the patient makes access difficult in case of resuscitation, and therefore the role of the anesthesiologist in the care of such patients is crucial in the perioperative period. Before the operation was performed, the patient was assessed by anaesthesiologist and was assigned ASA III (American Society of Anesthesiologists score is a subjective assessment of a patient's overall health that is based on five classes) due to severe congenital malformation with orthopedic and urological comorbidities. Apart from that, her physical condition was overall satisfying. Past anesthetic procedures have passed without complications. However, due to her condition that affects total body surface and volume distribution, preoperative preparation and assessment is of key importance in inducing and maintaining safe anesthesia as well as postoperative care. The patient was pre-medicated by midazolam and accompanied to the operating room by medical staff. After checking the identity and documentation, non-invasive blood pressure (NIBP) was placed on the right leg appendage. Furthermore, electrocardiogram (ECG) electrodes were placed on the chest as part of standard anaesthetic monitoring as well as pulse oximeter for oxygen saturation (SpO2) measurement on the right ear. Standard monitoring was extended by bispectral index monitoring (BIS). Initially, the patient was cardiopulmonary compensated with preoperative blood pressure of 120/60 mmHg and heart rate of 70 beats per minute. Her oxygen saturation was 100% without oxygen therapy. Initial laboratory results including coagulation tests were in reference range, and no noted coagulation abnormalities were noted. Before the surgery started, a venous line had to be placed, but due to the lack of peripheral venous access, the external jugular vein on the neck was cannulated. After careful denitrogenation, general endotracheal anesthesia was induced, and airway secured by flexible endotracheal tube number 5.5 with cuff. After induction, a urinary catheter and nasogastric tube were inserted. Drugs used for induction were midazolam, propofol, sufentanil and vecuronium. The patient was put on mechanical ventilation in volume mode. Anesthesia was maintained with propofol continuously on perfusor by rate of 160 mg/h with vecuronium fractionated to keep the patient's muscles relaxed. The patient prophylactically received cefazolin 400 mg intravenously (iv) to prevent infection and tranexamic acid 250 mg iv to reduce intraoperative bleeding. Surgery was performed in the prone position. During the procedure, the patient was tachycardic up to 170 beats per minute, and hypotensive down to 80/40 mmHg. Volume was compensated with crystalloids and erythrocyte concentrate. She has lost

about 350 ml of blood. Intraoperative autotransfusion device was used and by so, 75 ml of blood was returned into the patient's bloodstream. Postoperatively, the patient was intubated, sedated and transported to the pediatric Intensive care unit (ICU). Soon after arrival, she was extubated, and during the further course of her stay was hemodynamically and respiratorily stable. The patient has received two doses of erythrocyte concentrate, as well as the necessary analgesia and sedation. She was parenterally hydrated with a glucose-electrolyte solution, but oral intake of food and liquids has not been started. Hemodynamically and respiratorily stable, she was transferred to the Orthopedic ICU, where cardiorespiratory monitoring was maintained regularly, antibiotic prophylaxis, analgesia and sedation were administered. The patient is furthermore transferred to the Ward, where she was bandaged regularly, and physical therapy was started under the supervision of physiotherapist. During her hospital stay, the patient started sitting in a wheelchair while the wound healing was satisfactory. The patient subjectively felt well and was discharged successfully from the hospital.

Discussion

There are currently no recommendations in literature regarding the optimum management of these patients. A variety of corrective surgical procedures may be undertaken for malformations associated with amelia and phocomelia. According to the definition of the Scoliosis Research Society, scoliosis is any lateral curvature of the spine in the frontal plane whose Cobb angle is greater than 10 degrees. There are a number of classifications of scoliosis. Considering the etiology, they can be divided into primary (idiopathic) and secondary. In childhood idiopathic are more common, and depending on the age of appearance, are divided into idiopathic infantile scoliosis that occurs in the first three years of life, juvenile scoliosis that occurs from the fourth to the tenth year life and adolescent that appear after the tenth year of life. The most common deformations (over 80% of deformities) are adolescent idiopathic scoliosis. (8). "Early onset scoliosis" is the term for scoliosis of idiopathic or secondary etiology that occurs before the age of ten. Severe curves and rapid progress can cause disorders of lung development, ventilation disorders, heart decompensation and respiratory insufficiency (9). In the treatment of early-onset scoliosis, a combination of conservative and, if necessary, surgical treatment is used. Conservative methods include physical therapy, plaster bandages, and orthotic hello traction. Surgical treatment for scoliosis is indicated, in general, for the curves exceeding 45 or 50 degrees by the Cobb's method. Larger the curve progress it is more difficult to treat. Posterior fusion with instrumentation has been a standard of the surgical treatment for scoliosis, while anterior instrumentation surgery has been a choice of treatment for the thoracolumbar and lumbar scoliosis because better correction can be obtained with shorter fusion levels (10). Due to the current advances in care, there are increasing numbers of adults affected by phocomelia that present with medical conditions that require emergency interventions (6). The challenges facing the anaesthesiologist, especially when dealing with patients who present phocomelia as part of a syndrome, are numerous and include difficulty in monitoring blood pressure. It may be impossible to measure the blood pressure non-invasively due to the absence of limbs or if they are attached to the trunk via very short appendages. Invasive blood pressure monitoring may itself be very challenging, too. The choice of arteries available is sometimes limited to the femoral and axillary arteries which may be aberrant in course and caliber causing major difficulties in accessing them. Alternative approaches for assessment of cardiovascular status to enable fluid management may need to be discussed, including non-invasive cardiac output monitoring. Furthermore, difficulties can occur regarding venous access; this can present a huge challenge as only central veins may be accessible. Early involvement of the anaesthetic team should be considered to secure central venous access or a peripherally inserted central catheter (PICC) in these patients, preferably on the day prior to the surgical procedure and in a high dependency environment. In these cases, ultrasound can be very helpful and is highly recommended (11). Also, intubation can be difficult due to the limited neck movement because of possible cervical spine abnormalities. Problems regarding regional techniques can be encountered as well due to the spinal deformities including anterior fusion of thoracic and lumbar vertebrae, spina bifida and scoliosis. It may make spinal or epidural anaesthesia more challenging (6). In cases where airway abnormalities are evident or when a difficult airway is expected, advanced planning, specialist instruments, and appropriately trained medical staff must be available. Also, some forms of phocomelia have been associated with micrognathia. The need for transfusion is usually dictated by the surgical procedure, but it should be mentioned that due to a reduced skeletal muscle mass which acts as a vascular reservoir, the need for blood transfusions may be increased (6). As already mentioned, appropriate preparation for difficult venous access must be addressed and central venous access should be considered.

Conclusion

Phocomelia is an extremely rare birth defect that falls under the broader category of congenital limb malformations, while amelia is the congenital absence of arms or legs or both. Perioperative anaesthetic management faces a variety of difficulties during treatment of these groups of patients. Some of the challenges include difficulty in monitoring blood pressure, venous access, difficult intubation and fluid management. Due to all of the above mentioned difficulties, preoperative preparation and assessment is of key importance in inducing and maintaining safe anesthesia as well as postoperative care.

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Conflict of interess
No conflict of interest to declare.

Ethical approval
For every elective and urgent procedure in our Hospital, it is required to obtain an informed consent form. The patient had signed the informed consent form and therefore gave the Hospital permission to perform procedures as well as use the data for scientific purposes with strong protection of personal information.

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