Anesthesia and perioperative management for a patient with Ullrich syndrome undergoing surgery for scoliosis

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Abstract: Ullrich syndrome is a rare congenital hypotonic-sclerotic muscular disorder in which affected children develop a slowly progressive scoliosis and contractures and limpness of joints. The disease causes increasingly invalidating contractures and hardening of the muscles of the neck and trunk. While this neuromuscular type of scoliosis is progressive, patients rarely attain the point of surgery due to their compromised general medical condition. This may explain the current lack of outcome data and the paucity of information on perioperative management for patients with Ullrich syndrome undergoing major surgery. The purpose of this report was therefore to describe our first experience with the perioperative and anesthetic management of a 15-year old boy presenting with Ullrich syndrome and a secondary invalidating scoliosis. The specific challenges of this condition characterized by severe restrictive lung disease and a challenging airway abnormality are discussed.

Key words: Ullrich; hypotonic-sclerotic muscular dystrophy; scoliosis; perioperative management.

INTRODUCTION

In 1930 Ullrich described two patients with congenital muscular dystrophy and defined the disorder as sclero-atomic muscular dystrophy. He reported clinical manifestations such as proximal joint contractures, striking distal hyperextensibility, protruding calcanei, hyperhydrosis, delayed motor developmental milestones (many patients have never been able to walk), but normal intelligence (16, 17). Other common features include congenital hip dislocation, follicular hyperkeratosis, a round face, prominent ears, soft velvety skin and abnormal scarring. Clinical signs of weakness and wasting are generalized and slowly progressive, and are associated with recurrent respiratory tract infections. Most patients die of respiratory failure between their first and third decade. This type of congenital muscular dystrophy is part of the spectrum of collagen type VI-related disorders caused by mutations in the genes COL6A1, COL6A2 and COL6A3 on chromosomes 2 and 21. The majority of cases is inherited in an autosomal recessive fashion. Histology of muscle biopsy shows remarkable variation in fiber size with hypertrophy and hypotrophy. Fiber necrosis is rare and there is a clear increase of endomysial connective tissue. Immunohistochemistry with collagen VI antibodies displays severe reduction or absence of the protein in muscle and cultured fibroblasts (6, 8-10, 12).

In the presence of Ullrich disease the development of a neuromuscular scoliosis may occur. We hereby describe the perioperative course of a 15-year old patient who presented with Ullrich’s syndrome and underwent surgery for treatment of scoliosis. Patients with neuromuscular types of scoliosis are generally considered at high risk for postoperative morbidity and complications, especially those with impaired lung function. The authors wish to describe their thorough preoperative assessment and multidisciplinary perioperative management that allowed them to guide this patient safely through the surgical procedure and perioperative episode.
CASE REPORT

A 15-year old boy with a congenital muscular dystrophy, diagnosed as Ullrich’s syndrome, presents with a disabling secondary neuromuscular scoliosis. His medical record revealed reduced intrauterine movements and a reduced gain in weight during pregnancy. After birth there was an extreme hypotonia. At the age of one year he was able to sit, but was never able to crawl or walk. The diagnosis of Ullrich’s muscular dystrophy was confirmed by muscle biopsy revealing abnormalities in collagen fibers IV and VI.

This patient underwent several surgical procedures under general anesthesia in the past: embolization of a persistant ductus arteriosus at the age of 2 and several orthopedic procedures (elongation of muscular tendons and surgery for femoral fracture), which were tolerated well.

More recently he presented to the pediatric orthopedic clinic with a C-shaped progressive scoliosis, interfering with his sitting balance and respiratory function. At that time, his orthopedic treatment consisted of adequate orthotic adaptation of his seating. However, due to progressive loss of balance in the frontal as well as the sagittal balance, further conservative management became impractical and resulted in worsening of the respiratory function. Thorax expansion for breathing had become only possible when this patient was leaning forward and supported himself on both elbows. Surgery aimed to stop a potential further progression of the spinal curve and to restore mainly the sagittal balance.

Preoperatively he underwent multidisciplinary assessment and counseling (pediatric, orthopedic surgeon, anesthesiologist, pulmonologist, neurologist, cardiologist and intensivist). The possible postoperative respiratory problems were discussed with the patient and his parents and the decision was made to wean the patient in intensive care.

The preoperative X-rays of the spine showed a lumbar curve with a sinistroconvex lumbar curve measuring 77°, without correction on the traction nor bending films. There was a co existence of unilateral painless subluxation of the right hip (Reimers 75%). There was no pelvic obliquity between the fifth lumbar vertebra and the pelvis.

He had an inappropriate staturoponderal evolution weighing 24 kilograms (height 155 cm) with remarkable intellectual capacities. He presented with weak mimic in facial muscles, weak force in the upper extremities and both lower extremities could not be moved against gravity. There was a generalized amyotrophy.

He had a severe progressive restrictive lung disease due to his thoracic deformation. There was no oxygenation problem, preoperative pulseoxymetry showed an oxygen saturation of 100% at room-air. At the age of 13 non-invasive nocturnal ventilation (BiPAP) was initiated because of CO₂ retention. He had recurrent respiratory tract infections and elective surgery had to be postponed for one week because of an episode of bronchitis with sputa, treated with amoxicillin/clavulanate. On auscultation there was wheezing during inspiration and expiration over the left lung. Pulmonary function tests revealed a forced vital capacity (FVC) of 770 ml (24% of predicted value), a forced expiratory volume in 1 second (FEV1) also 24% of predicted value and a Tiffeneau index of 84%. The preoperative hematocrit was 43.9%. Coagulation parameters, ECG and echocardiography were normal. The patient took no medication and had no documented allergies.

After overnight fasting, a non-invasive blood pressure, three leads ECG, pulse-oxymetry and a BIS monitor were attached.

The patient was induced with a bolus of sufentanil (10 mcg) and propofol (80 mg). Because he was already on non-invasive nocturnal ventilation we expected to be able to get a free airway. Difficult intubation equipment was available in the operating room. After verifying free airway, we gave a bolus of cisatracurium (3 mg). Total intravenous anesthesia (TIVA) with propofol (100 mcg/kg/min) and intermittent boluses of sufentanil were used to maintain anesthesia.

Due to the rigid musculature of the mouth and the presence of a big tongue, neither the vocal cords nor epiglottis could be visualised by direct laryngoscopy. We could intubate the trachea through the nose using the fiberoptic bronchoscope and inserted a cuffed endotracheal tube n° 6.

Ventilation settings on the Dräger Zeus mechanical ventilator were volume controlled mechanical ventilation at autoflow (closed circuit) with a mixture of O₂/air (FiO₂ : 0,5), inspiratory time was set at 33%, tidal volume at 240 ml and respiratory rate at 14.

According to our standard policy for scoliosis surgery, nasogastric tube and oesophageal temperature probe were positioned. Body temperature of the patient was maintained using both a forced hot air blanket (Bair Hugger) and an IV fluid heating device (Fluido). An invasive arterial line was inserted in the right radial artery and a central
venous line (guided by ultrasound) was placed in the right internal jugular vein. An urethral catheter was inserted to monitor urinary output.

A red blood cell saving device (Cellsaver Electa, Dideco) was used during surgery. Cefazoline was used as antibiotic prophylaxis.

The surgery was performed in prone position with cushions placed under the chest and pelvis of the patient. We installed a preformed cushion under the forehead of the patient and therefore had to remove the BIS electrodes to prevent decubitus lesions.

Due to lack of respiratory reserve, it was decided not to perform an anterior release of the lumbar region despite its theoretical advantage of obtaining full spinal balance. A partial surgical correction was envisaged in order to improve sitting balance and to make further orthotic treatment feasible. A classic posterior approach to the spine in prone position was therefore carried out. A segmental instrumentation was performed using pedicle screw instrumentation from T2 towards L5 (Xia Stryker 4,5 mm) assisted by computer Navigation (Stryker) and image intensifier. The remaining muscle on the concave side was partially resected in order to get a release of the lumbar region of the spine. A partial reduction of the stiff lumbar curve could be obtained (lumbar curve reduced to 50°) and the sagittal balance could be restored. A derotation of the apex of the curve was not feasible due to stiffness of the curve and a poor bone quality.

The operative procedure took 6 hours, and blood loss was approximatively 400 ml (i.e. 25% of total circulating blood volume).

During surgery 3,5 liters of crystalloids (Hartmann solution) were administered. The washed content of the cell saving apparatus was returned to the patient (343 ml of washed RBC).

At the end of the procedure our ventilator settings were adjusted due to increased peak pressures (up to 38 cmH2O) and increased PaCO2. Tidal volume was reduced to 200 ml and respiratory rate was set at 20.

After the procedure a drop in blood pressure was seen after turning the patient in supine position. This was treated with additional fluid administration (400 ml of colloids).

The patient’s trachea remained intubated for weaning in the pediatric intensive care unit (PICU) as discussed with the patient and parents preoperatively.

There was minimal postoperative bleeding. During the first 16 hours Pressure Regulated Volume Control mode (Siemens ventilator) was applied, requiring only low ventilation pressures (PIP 15 mmHg/ PEEP 5 mmHg) at FiO2 of 0.3. After stopping the analgosedatives (morphine/midazolam) weaning was continued at Volume Support mode. There was an excellent gasexchange throughout. Twelve hours later his trachea was extubated in the presence of an anesthesiologist. After uneventful extubation continuous non-invasive ventilation (BiPAP) was applied using his home device, and room air only. The postoperative orthopedic recovery was uneventfull and did allow for an improved sitting position, with improved frontal and sagittal balance. The fixation was stable and did not need any further bracing. The patient could be positioned in upright sitting position when desired. He was discharged from the PICU on the 6th postoperative day and left the hospital uneventful on the 12th postoperative day after adjusting his seating orthoses.

**DISCUSSION**

This article describes the details of a multidisciplinary approach to the perioperative care of a patient with Ullrich disease presenting with a severe neuromuscular scoliosis. The management of the patient led to an uneventful recovery despite his poor general medical condition.

In several other medical centers the patient had repeatedly been denied surgical treatment because of the severely reduced lung function that can be considered as an excessively high risk perioperative risk. However, because further conservative treatment became impractical and resulted only in further deterioration of his respiratory status, the patient and his parents came to us with a clear call for help. Considering the potential benefit of scoliosis surgery and a relatively poor predictive value of preoperative lung function tests in this condition, we reevaluated the possibility of surgery. In selecting treatment options for this type of patient it is important to realise that the mental status and cognitive function of patients with Ullrich syndrome are completely normal. A thorough description of all treatment options with the patient and parents including detailed information with respect to risks and outcome of the procedure remains one of the best anxioalytic methods (7, 14).

Of particular interest to the anesthesiologist are the changes in musculature of the mouth and cervical region. All precautions for a difficult intubation scenario need to be present. Because the
operative management are therefore mandatory to preoperative assessment and multidisciplinary perioperative approach.

A second challenge was the choice of anesthetics. Volatile anesthetics are associated with an increased risk of rhabdomyolysis as a result of toxic effects on diseased muscle with exacerbation of breakdown of frail membranes (1, 5). Nevertheless, we preferred to use TIVA guided by BIS monitoring. The potential side effect of this technique is the development of propofol infusion syndrome causing hyperkalemia, metabolic acidosis, rhabdomyolysis and myocardial failure (3, 4, 11). However this is usually associated with high doses of propofol (> 4 mg/kg/hour) administered for a prolonged period of time (> 48 hours) although recent case reports suggest metabolic lactic acidosis with shorter infusions of propofol (11). Although we planned to tailor propofol dosing based on BIS monitoring, the electrodes had to be removed once the patient was turned in prone position, to avoid skin lacerations. For sedation in the postoperative phase, we switched to midazolam/morphine to avoid an excessive propofol load.

We used a non-depolarizing neuromuscular blocking agent, because succinylcholine is contraindicated. Succinylcholine may damage the muscle membrane and release intracellular potassium.

Despite the lack of a general consensus it has been suggested that the median survival in patients with similar type of muscular dystrophy (Duchenne) may improve with the combined use of nocturnal ventilation and spinal surgery (2). This statement needs to be balanced with the fact that patients with neuromuscular scoliosis are considered to be at high risk for postoperative morbidity and complications due to their underlying compromised respiratory condition. Especially those with a low FVC and FEV1, are at risk for need of prolonged postoperative mechanical ventilation (15). The main reasons for prolonged ventilatory support are: a suboptimal result of surgery, hypoventilation due to pain, effects of residual anesthetic drugs and muscle weakness due to immobility (19). Careful preoperative assessment and multidisciplinary perioperative management are therefore mandatory to limit postoperative morbidity, when selecting major spinal surgery in pediatric patients with severely decreased vital capacity (13, 18).

In a climate of growing public awareness, patients and their parents deserve extensive preoperative counseling regarding risks involved in anesthesia, surgery and postoperative course. This can only be accomplished by means of a multidisciplinary perioperative approach.

References


