

Perioperative care of a child with Ullrich congenital muscular dystrophy during posterior spinal fusion

Martin DP, MD, Department of Anesthesiology and Pain Medicine, Nationwide Children's Hospital, Columbus, Ohio, USA

Tobias JD, MD, Chairman, Department of Anesthesiology and Pain Medicine, Nationwide Children's Hospital
Professor of Anesthesiology and Pediatrics, The Ohio State University, Columbus, Ohio, USA

Warhadpande S, BS, The Ohio State University School of Medicine, Columbus, Ohio, USA

Beebe A, MD, Department of Orthopedic Surgery, Nationwide Children's Hospital, Columbus, Ohio, USA

Klamar J, MD, Department of Orthopedic Surgery, Nationwide Children's Hospital, Columbus, Ohio, USA

Correspondence to: Joseph Tobias, e-mail: joseph.tobias@nationwidechildrens.org

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Abstract

Ullrich congenital muscular dystrophy (UCMD) is one of a group of disorders known as congenital muscular dystrophies. Severe hypotonia and early diaphragmatic involvement may lead to respiratory failure early in the disease process. We present the case of a nine-year-old with UCMD who required operative intervention for progressive scoliosis. In these patients, anaesthetic issues relate to difficulties with endotracheal intubation, as well as the potential for postoperative respiratory failure, given early diaphragmatic involvement. As with other types of muscular dystrophy, succinylcholine is absolutely contraindicated, while a prolonged effect may be seen following routine doses of nondepolarising neuromuscular blocking agents. Additional perioperative concerns relate to the surgical procedure primarily, including tailoring the intraoperative anaesthetic to facilitate neurophysiological monitoring, as well as the use of techniques to limit intraoperative blood loss. The perioperative management of patients with UCMD is discussed and options for intraoperative anaesthetic care are reviewed.

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Introduction

Ullrich congenital muscular dystrophy (UCMD) was first reported in 1930 by Otto Ullrich, who cared for two children with proximal joint contractures and severe distal joint hyperextensibility. The disorder was subsequently named congenital atonic-sclerotic muscular dystrophy, but later became known as Ullrich congenital muscular dystrophy. UCMD is inherited as an autosomal recessive trait, that usually manifests shortly after birth with hypotonia. Subsequently, the classical phenotype develops, including axial muscle contractures and distal joint hyperlaxity. Additional clinical features include a high-arched palate, protuberant calcanei and rigid spine syndrome. Despite the extensive and progressive nature of the muscular involvement, intelligence and cognitive development are normal.¹ There is an early and progressive deterioration of diaphragmatic function in the majority of patients. This leads to respiratory insufficiency and the need for respiratory support.

The primary defect in UCMD is the deficiency of collagen type VI, which is present in the extracellular space that surrounds the muscle fibres and acts as an anchor between

the basement membrane and the underlying endomysium.² Deletion of the COL6A1 gene in UCMD results in the deficiency of collagen type VI. This causes congenital muscular weakness, proximal joint contractures, distal joint hyperextensibility, spinal rigidity and scoliosis.³ Given the associated involvement of several organ systems, as well as the various linked orthopaedic manifestations, surgical interventions and anaesthetic care may be required in patients with UCMD.^{4,5} We present the case of a nine-year-old girl with UCMD who required perioperative care during posterior spinal fusion for scoliosis. Perioperative care of these patients is discussed.

Case report

Institutional board review and approval are not required for single case reports at Nationwide Children's Hospital (Columbus, Ohio, USA). The patient was a 32-kg, nine-year-old girl who presented with neuromuscular scoliosis that was secondary to UCMD. The upper thoracic curve (T1-T3) measured 57 degrees. The patient's past medical history revealed that UCMD was diagnosed in infancy when she presented with hypotonia, poor head control and failure to walk. She also had a history of gastrooesophageal reflux

that was treated with omeprazole, and poor oral intake that required gastrostomy tube feedings predominantly.

During an episode of pneumonia 8-9 months prior to the scheduled surgery, the patient required endotracheal intubation and mechanical ventilation, followed by tracheostomy and home mechanical ventilation. At that time, her trachea could not be intubated via standard direct laryngoscopy and indirect video laryngoscopy with the Glidescope® was used in the paediatric intensive care unit by the paediatric anaesthesiology staff.

When she presented, she was being ventilated at home through a 5-mm uncuffed Shiley® tracheostomy tube. Her parents reported occasional ventilator alarms because of low-minute ventilation. Home ventilator settings included synchronised intermittent mandatory ventilation with a rate of 14 breaths/minute, a peak inflating pressure of 20 cmH₂O, positive-end expiratory pressure of 8 cmH₂O, pressure support of 10 cmH₂O and fraction of inspired oxygen (FiO₂) of 0.28. Her latest chest radiograph showed minimal atelectasis of the right lung field that had improved from a previous film. Past surgical history included a muscle biopsy, placement of a gastrostomy tube and a tracheostomy. A physical examination revealed no acute distress. The tracheostomy tube was in place. There was moderate micrognathia with limited mouth opening (2 cm). Her lung fields were clear to auscultation. There were contractures of the elbows, wrists, hips and knees, with decreased strength of all muscles (two out of five). Medications included omeprazole (20 mg twice a day) via the gastrostomy tube, and albuterol (2.5 mg twice a day) administered via a high-flow nebuliser. Preoperative laboratory evaluation revealed haemoglobin 14 gm/dl, haematocrit 43.4% and a platelet count of 450 000/mm³. The prothrombin time was 14.5 seconds, the international normalised ratio was 1.13 and the activated partial thromboplastin time was 30.

On the day of the surgery, the patient was held nil per os for six hours, except for her usual morning dose of omeprazole. A peripheral intravenous cannula was placed and premedication was provided by midazolam (0.05 mg/kg). The patient was transported to the operating room and routine American Society of Anesthesiologists monitors were placed. Preanaesthetic oxygen saturation was 96% and blood pressure, 127/68 mmHg. The patient was preoxygenated with 100% oxygen via the tracheostomy and anaesthesia was induced with propofol (2 mg/kg) and fentanyl (3 µg/kg). Neuromuscular blockade was achieved with a single dose of rocuronium (0.3 mg/kg). Anaesthesia was then provided with desflurane (exhaled concentration of 3-4%) titrated to maintain the bispectral index at 40-60.

Following anaesthetic induction, a second large-gauge peripheral intravenous cannula and a radial arterial cannula were placed. Remifentanyl was added to the maintenance anaesthesia regimen, titrated from 0.1-

0.4 µg/kg/minute to maintain the mean arterial pressure at 55-65 mmHg. Tranexamic acid was administered as a bolus dose of 100 mg/kg, followed by an infusion of 10 mg/kg/hour. Perioperative antibiotic prophylaxis was provided by clindamycin, given a penicillin allergy. The Shiley® tracheostomy tube was removed and replaced by a 4.5-mm cuffed, reinforced endotracheal tube that was secured via the existing tracheostomy. Monitoring leads were placed for motor and somatosensory-evoked potentials. The patient was positioned prone on the Jackson table. The surgical procedure, which included posterior spinal fusion with instrumentation (T2-S2), pelvic fixation and allograft bone grafting, lasted 5.5 hours. Estimated blood loss was 1 700 ml. Intraoperative fluids included three units of packed red blood cells, 800 ml of 5% albumin and 2 000 ml of lactated Ringer's. Intraoperatively, three doses of sodium bicarbonate (1 mEq/kg) and three doses of calcium chloride (10 mg/kg) were administered to treat metabolic derangements. The metabolic acidosis was judged to be dilutional as the plasma lactate concentration was not elevated. Postoperatively, the patient received one transfusion of fresh frozen plasma and platelets to correct coagulation function. The remainder of her postoperative course was uncomplicated and she was discharged on postoperative day seven.

Discussion

The congenital muscular dystrophies are a heterogeneous group of diseases that affect various components of skeletal muscle.⁶ These disorders, which involve the muscular architecture (glycosylation process or structural components of the extracellular matrix), include UCMD, merosin-deficient congenital muscular dystrophy and rigid spine syndrome. The congenital muscular dystrophies present with similar clinical findings in utero during the first few months of life with decreased intrauterine movement, hypotonia and the delayed achievement of motor milestones, including decreased movement in utero. Despite the similarities, there are specific signs and symptoms that may suggest the diagnosis of UCMD, including spinal rigidity, hyperlaxity of the joints of the hands and feet and early diaphragmatic involvement, leading to respiratory failure.

Of significant concern during the perioperative care of such patients is the potential for difficult endotracheal intubation, as well as concerns about respiratory failure. In these patients, difficult endotracheal intubation may relate to limited mouth opening, as well as limited cervical spine movement because of the involvement of the musculature of the head and neck. Both of the previous reports of anaesthesia for patients with UCMD mention difficulties with endotracheal intubation.^{4,5} Airway concerns were not an issue in our patient, given the presence of a tracheostomy. However, difficulties with endotracheal intubation were demonstrated by her previously requiring endotracheal intubation via

indirect video laryngoscopy. Although the previous reports suggest that bag-valve-mask ventilation may be normal in patients with UCMD,^{4,5} we recommend that spontaneous ventilation is maintained during anaesthetic induction until the ability to provide effective bag-valve-mask ventilation is demonstrated. Additionally, the appropriate equipment that is needed to deal with the difficult airway should be readily available.⁷

Although postoperative mechanical ventilation was necessary in our patient, given her baseline respiratory failure and chronic home mechanical ventilation, preoperative preparation and postoperative attention to respiratory function are recommended in patients with UCMD. In patients who have not yet progressed to requiring chronic ventilator support, the residual effects of anaesthetic agents may impact on upper airway control and diaphragmatic function, thereby resulting in perioperative respiratory failure. Whenever feasible, short-acting agents whose effects dissipate rapidly, such as remifentanyl or desflurane, may be beneficial. Regardless of the selected agents, postoperative monitoring of respiratory function is suggested. As appropriate, based on the surgical procedure, regional anaesthesia may be chosen instead of general anaesthesia, or as an adjunct to provide postoperative analgesia, and thereby minimise the perioperative effects of opioids.⁵ Preoperative preparation should include aggressive treatment of respiratory infections and use of techniques, such as incentive spirometry or other noninvasive techniques of respiratory support, to prevent atelectasis and further deterioration in respiratory function.^{8,9} In our experience, the postoperative use of noninvasive ventilation, such as bilevel positive airway support, may facilitate postoperative tracheal extubation in patients with altered respiratory function.

Following anaesthetic induction and endotracheal intubation, the appropriate agents for maintenance anaesthesia need to be chosen. In our patient, the agents for maintenance anaesthesia were somewhat dictated by the use of neurophysiological monitoring of spinal cord integrity, including motor and somatosensory-evoked potential monitoring.^{10,11} Based on our usual practice, this included a combination of the inhalational anaesthetic agent, desflurane, and a continuous infusion of remifentanyl. These agents were chosen based on their ability to provide effective analgesia and amnesia without the use of a neuromuscular agent, to allow for rapid awakening and to provide a stable baseline anaesthetic to facilitate neurophysiological monitoring.

Although we chose to use the volatile agent, desflurane, the use of these agents is considered to be controversial by some investigators who have suggested that a total intravenous anaesthetic technique should be used.^{12,13} However, it must be noted that the majority of this

information relates to experience with Duchenne and Becker types of muscular dystrophy. There are no available data on many of the less common forms of muscular dystrophies, including UCMD. As previously thought, these patients are not at increased risk of malignant hyperthermia. However, the prolonged use of a volatile anaesthetic agent may result in disease-related cardiac complications, or rarely, a disorder that clinically resembles malignant hyperthermia which relates to destabilisation of the sarcolemma and causes rhabdomyolysis and hyperkalaemia.¹⁴⁻¹⁷ This latter complication may also occur postoperatively. As opposed to patients with Duchenne or Becker muscular dystrophy, no cardiac involvement is noted in patients with UCMD. Also, there have been no reports that hyperkalaemia relates to the use of volatile anaesthetic agents.¹⁸

Another issue regarding endotracheal intubation and maintenance anaesthesia in patients with UCMD is the choice of neuromuscular blocking agent. Given the risks of rhabdomyolysis, hyperkalaemia and cardiac arrest with succinylcholine, this agent should never be used in patients with muscular dystrophies.^{19,20} Conversely, although nondepolarising neuromuscular blocking drugs (NMBDs) are safe in patients with various muscular dystrophies, a prolonged effect can be expected, even with intermediate-acting agents, such as atracurium, rocuronium or vecuronium.^{19,20} Prior to their withdrawal, rapacuronium and mivacurium, two short-acting nondepolarising NMBAs, offer the ability to achieve complete neuromuscular blockade to allow for endotracheal intubation with an acceptable recovery time, even for briefer procedures.^{21,22} Alternatively, rapid-sequence intubation can be accomplished using a combination of propofol and remifentanyl without a neuromuscular blocking agent.^{5,23,24} Given that our patient was already receiving chronic mechanical ventilation at home, the plan was to continue mechanical ventilation postoperatively. Therefore, concerns about residual neuromuscular blockade were not clinically relevant. Additionally, given the anticipated length of the procedure (5-6 hours) and the small dose of rocuronium that was administered (0.3 mg/kg), the likelihood of residual neuromuscular blockade, even in a patient with a muscular dystrophy, was limited. The other possibility was the use of sugammadex to eliminate the potential for residual neuromuscular blockade and to ensure complete return of skeletal muscle strength. However, sugammadex is not commercially available in the USA.

Additional perioperative concerns about our patient related to the surgical procedure, rather than the underlying disease process itself, including blood loss. As illustrated by the most recent work from the Perioperative Cardiac Arrest registry, blood loss is the most usual aetiology of perioperative cardiac arrest, while the most commonly implicated surgical procedures are craniofacial reconstruction and posterior spinal fusion.²⁴ Additionally, blood loss is greater

in patients with neuromuscular scoliosis vs. idiopathic varieties.²⁵ As such, appropriate intravenous access, as well as ready access to blood and blood products during the procedure, is suggested. Furthermore, consideration should be given to employing techniques that may effectively limit intraoperative blood loss.²⁶ In our patient, controlled hypotension, intraoperative blood salvage and the administration of the antifibrinolytic agent, tranexamic acid, were employed.²⁷

Conclusion

In conclusion, we present the case of a nine-year-old girl with UCDM who required perioperative care during a posterior spinal fusion. In these patients, anaesthetic issues relate to difficulties with endotracheal intubation primarily, as well as the potential for postoperative respiratory failure, given early diaphragmatic involvement. As with other types of muscular dystrophy, succinylcholine is absolutely contraindicated, while a prolonged effect may be seen following routine doses of nondepolarising agents. Unlike other types of muscular dystrophy, cardiac involvement has not been reported in patients with UCMD. Additional perioperative concerns related to the surgical procedure mainly, including tailoring the intraoperative anaesthetic to facilitate neurophysiological monitoring, as well as the use of techniques to limit intraoperative blood loss.

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