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General Anesthesia for a Patient with Centronuclear (Myotubular) Myopathy

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CENTRONUCLEAR myopathy[1-4] is a rare congenital myopathy characterized by progressive muscle weakness. We report a case of general anesthesia for a patient with this.

Case Report

A 32-yr-old woman with centronuclear myopathy was scheduled for bilateral arthroplasties for treatment of ankylosis of the temporomandibular joints. For several years before surgery, she had progressive ankylosis with decreasing range of motion of her mandible. Oral intake was possible only through a small straw, and oropharyngeal secretions could be cleared only with a small soft suction catheter.

The diagnosis of centronuclear myopathy was made at 6 yr of age and was reported in the literature as one of the first examples of this myopathy.[5] Because of the disease she was wheelchair-bound at an early age. Subsequently restrictive lung disease and asthma developed with multiple episodes of bronchitis and pneumonia necessitating hospital admission. Although pulmonary function testing indicated severe restrictive lung disease, the arterial blood gas levels were normal. The patient had sleep apnea necessitating nasal bilevel pressure-assisted ventilation at night. A sleep study performed previously in the year while undergoing nasal bilevel pressure-assisted ventilation showed multiple oxygen desaturations throughout the night, with 53 events below 90% to as low as 68%. She also had a history of gastroesophageal reflux, but was without cardiac, hepatic, renal or other neuromuscular disorders. At the time of admission, medications included theophylline, terfenadine, and prednisone. She was allergic to sulfa, dust, pig hair, cow hide, and tree pollen.

Surgical history included correction of strabismus at 4 yr of age, bilateral ptosis repair at 6 yr of age, and bilateral Achilles tendon release at 12 yr of age, all performed uneventfully during general anesthesia. One year before admission, the patient underwent temporomandibular joint surgery at another institution, where she required emergent oral intubation for apnea after premedication with droperidol and fentanyl.

Physical examination revealed a thin woman in a wheelchair who became mildly short of breath while speaking. She was unable to open her mouth. Her chest was scoliotic with breath sounds decreased bilaterally. Wheezes were absent. Muscle strength was 3/5 in all extremities.

Before surgery, the anesthesia machine was flushed with oxygen. At arrival in the operating room, glycopyrrolate was administered, and airway anesthesia was obtained using topical lidocaine and percutaneous superior laryngeal nerve blocks. After uneventful nasal fiberoptic intubation with a 6.0-mm nasal R.A.E. endotracheal tube, general anesthesia was induced with propofol and maintained with a propofol infusion, nitrous oxide, and oxygen. No additional intravenous medications were administered at any time, except for ketorolac 1 h before emergence.

Bilateral coronoidectomies with bilateral arthroplasty with eminectomies were performed without difficulty. The surgical sites were infiltrated with bupivacaine by the surgeon. The patient awoke and was transported to the postanesthesia care unit while breathing spontaneously. After uneventful extubation, she was placed on nasal bilevel pressure-assisted ventilation at her usual settings. The remainder of her postoperative course was unremarkable, and she was discharged on the third postoperative day.

Discussion

Centronuclear myopathy was first described in 1966.[6] It presents in early childhood with slowly progressive weakness of the extraocular, facial, neck, and limb muscles.[1-4] Because of the histologic resemblance of the diseased muscle to fetal myotubes, this disorder originally was called myotubular myopathy. However, the

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similarity between fetal myotubes and diseased muscle tissue is not complete, and the term centronuclear myopathy, which refers to the large number of centrally placed nuclei in specimens of diseased muscle, is preferred. As more cases were reported, it became clear that there was considerable heterogeneity with respect to presentation, severity, histology, and inheritance. The reported cases included several with severe X-linked disease for which the term myotubular myopathy is used by some authors.[1] Although the pattern of inheritance helps to distinguish the different variants,[7] the clinical presentation of each is similar enough to consider as a single disease.

Autosomal dominant and autosomal recessive forms of centronuclear myopathy often are divided, although imperfectly, with respect to the age of onset.[1] The early-onset form is the most common, follows an autosomal recessive pattern of inheritance, and can be seen in white and black patients. It may present at birth and may be severe enough to produce respiratory distress at that time. It is slowly progressive, but most patients die or are wheelchair-bound by the second or third decade of life. The development of scoliosis with restrictive lung disease is the most important physiologic manifestation of the disease's progress.[8] As with our patient, the association of ptosis and strabismus may increase the likelihood of surgical procedures during childhood. Seizures, some degree of mental retardation, psychosis, or other central nervous system disorders will develop in many children. Congenital lesions of the heart and cardiomyopathy occasionally are described.[2] A high, arched palate may be present.

Late-onset centronuclear myopathy follows an autosomal dominant pattern. It usually becomes apparent by the third decade of life.[1-4] The progress of the late-onset form is slower than that of the early-onset form but can still leave some patients wheelchair-bound by the sixth decade of life. The degree of defect can become worse during pregnancy.

In centronuclear myopathy, serum creatine kinase concentrations usually are normal, although mild elevations are sometimes seen with the early-onset form of the disease.[1] Changes consistent with myopathy are observed on the electromyogram. However, conduction velocities of the motor nerves are normal. Electroencephalographic abnormalities have been noted in several patients with the early-onset form of the disease before clinically apparent seizure activity begins.

Some authors use the term myotubular myopathy for the severe nonprogressive X-linked form of centronuclear myopathy in which few of the boys born with this disorder are able to overcome the marked respiratory distress present at birth.[1-4] Although males are severely affected, asymptomatic mothers have been identified using muscle biopsy. Micrognathia is one of several dysmorphic features that may be present in these patients. Serum creatine kinase concentrations usually are normal. The results of the electromyographic examination are consistent with myopathy, and conduction along the motor nerves is normal. In patients with X-linked myotubular myopathy, molecular biologic techniques have shown mutations in the gene encoding the protein myotubularin.[9,10]

In this patient, several issues directly related to the myopathy influenced the choice of an anesthetic plan. These were the potential for muscle relaxants to interact with the diseased muscle tissue[11] and the possibility of triggering malignant hyperthermia.[12] Muscle relaxation was avoided and a nontriggering anesthetic was used. The severe restrictive lung disease, sleep apnea, difficult airway secondary to ankylosis of the temporomandibular joints, history of bronchospasm, and gastroesophageal reflux also were of concern.

We conclude that patients with centronuclear myopathy can be treated successfully using an anesthetic plan that directly addresses the presence of the underlying myopathy and the associated decrements in physiologic function.

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Transfusion-related Acute Lung Injury (TRALI) Complicating Colectomy for Ulcerative Colitis

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TRANSFUSION-RELATED acute lung injury (TRALI) is a life-threatening complication that presents as adult respiratory distress syndrome after recent blood transfusion. The diagnosis may not be immediately obvious, especially if another systemic illness complicates the clinical picture. The immune basis of this transfusion reaction usually is caused by an incompatibility of recipient white cells and donor human leukocyte antigen (HLA) antibodies. Therefore, routine transfusion reaction evaluations, which explore erythrocyte incompatibilities, will fail to confirm the diagnosis. We report a case of TRALI, complicating colectomy for active ulcerative colitis. Postreaction immunologic evaluation clarified the pathogenesis of this event. Our findings point to the role of donor and recipient mediators. When transfusing blood during active inflammatory disease, minimizing potential white blood cell and serum interactions may be considered.

Case Report

A 12-yr-old, 53.6-kg girl with ulcerative colitis, which was unresponsive to medical therapy, was scheduled for colectomy. Preoperative course was notable for febrile episodes coincident with bowel preparation and for a hemoglobin level of 7.7 g/dl. The child received 500 cc irradiated, packed erythrocytes the evening before surgery, with no posttransfusion complications.

Induction and maintenance of general anesthesia with pentothal, fentanyl, hydromorphone, vecuronium, and isoflurane were uneventful. Approximately 5 h after the start of the case, the patient's hemoglobin level decreased from 8.1 to 6.1 g/dl. A transfusion of whole blood was administered. After completion of the blood transfusion, the patient's peak airway pressure suddenly increased from 23 to 45 cm water, and copious secretions were noted issuing from the endotracheal tube. Pulmonary auscultation revealed distant bilateral breath sounds, but no expiratory wheezes. The endotracheal tube was suctioned repeatedly for large amounts of clear, frothy fluid. Arterial blood gas revealed the following: *p*H: 7.21; P_{O2}: 69 mmHg; P_{CO_2}: 66 mmHg; HCO_3^-, 25.2 meq/L; base excess: 3 meq/L. The patient was administered furosemide, hydrocortisone, and diphenhydramine. Oxygen (100%) was administered with 10 cm water positive-end expiratory pressure. Systolic blood pressure decreased to 70 mmHg, necessitating several bolus doses of epinephrine to restore a normal blood pressure. Central venous cannulation was performed, revealing a central venous pressure of 14 mmHg. Chest radiography showed diffuse bilateral pulmonary infiltrates.

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