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CLINICAL REPORT

Anesthesia in a patient with mucopolysaccharidosis type VI (Maroteaux–Lamy syndrome)

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Abstract We report a case of anesthesia during surgery to enlarge the foramen magnum in a pediatric patient with an extremely rare form of mucopolysaccharidosis type VI (Maroteaux–Lamy syndrome). Airway control was unexpectedly easy, and intraoperative anesthetic management with total intravenous anesthesia went smoothly. However, the disease is progressive, with no guarantee that future anesthetic management of this patient will remain easy. If repeated surgery is required, thorough testing should be conducted over time to assess both airway and systemic complications. Nevertheless, we found that safe anesthetic management of affected patients is possible with anesthetics currently used in a clinical setting.

Keywords Mucopolysaccharidosis · Maroteaux–Lamy syndrome · Difficult airway · Pediatric anesthesia

Introduction

Mucopolysaccharidosis is a hereditary disease in which a deficiency of lysosomal enzymes that break down mucopolysaccharides causes the latter to accumulate throughout the body. The systemic deposition of mucopolysaccharides causes disorders of various organs. Central nervous system disturbances such as compressive myelopathy caused by dural and ligamentous thickening, and hydrocephaly caused by thickening of the arachnoid villi, have been reported. Accumulation of mucopolysaccharides in the heart causes cardiac complications such as sinus tachycardia, atrial dilatation, valvular disorders, myocarditis, and endocarditis. Furthermore, airway complications caused by scoliosis, atlantoaxial dislocation, and upper airway obstruction from mucopolysaccharide accumulation have also been reported. These conditions require attention during anesthetic management [1, 2]. Depending on the specific type of lysosomal enzyme deficiency, mucopolysaccharidosis can be classified into six subtypes, with mucopolysaccharidosis type VI (Maroteaux–Lamy syndrome) being extremely rare. To the best of our knowledge, only one other report has described anesthesia in a type VI patient [3]. However, the reported patient was 12 years old, and there was no description of anesthesia in early childhood of a type VI patient. Herein, we report anesthetic management for a patient with mucopolysaccharidosis type VI.

Case presentation

The patient was a 9-month-old boy (height 77.5 cm, body weight 9.5 kg), who had been delivered vaginally in gestational week 39 and weighed 3,222 g at birth (Apgar scores, 8/1 and 9/5 min). No notable problem was observed during the perinatal period. However, medical examinations conducted by an orthopedic surgeon at 1 month after birth showed limited abduction in flexion as a result of bilateral hip contracture and an irregular margin of the femoral head, and the patient was followed. At 6 months old, thorough medical examinations were conducted to ascertain the cause of bilateral leg dysfunction, during

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which enlarged head circumference, scoliosis, and lumbar vertebral deformation were noted. Various enzyme activities were tested, with only a low level of leukocyte arylsulfatase B found. Therefore, the patient was diagnosed with mucopolysaccharidosis type VI.

From around 8 months old, reduced mobility of the right upper and lower extremities became marked, and head computed tomography (CT) findings showed hydrocephaly. At another hospital, a ventriculoperitoneal shunt (VP shunt) was used to control cerebral pressure, and enzyme replacement therapy was initiated for the primary disease. At the time of VP shunt surgery, the patient was 8 months old (height 74 cm, body weight 9 kg). Anesthesia was introduced by slow induction with 8% sevoflurane, and tracheal intubation was performed easily. For anesthetic maintenance, air, oxygen, and 1.5% sevoflurane were used, with fentanyl given. The surgery was completed in 1 h and 40 min, and the period of anesthesia was 3 h, with no difficulties encountered. Thereafter, magnetic resonance imaging (MRI) was performed with sedation by triclofos sodium syrup. During MRI study, his spontaneous breathing was stable. MRI confirmed spinal compression caused by hypoplasia of the first cervical vertebra (Fig. 1), and hydrocephaly and poor movement of the right upper and lower extremities were attributed to spinal compression at the first cervical vertebra. Thus, the patient was referred to the Department of Pediatric Neurosurgery at our center to undergo surgical decompression to enlarge the foramen magnum.

External findings included increased head circumference, mucopolysaccharidosis-like facial appearance, swollen hands and feet, and right convex scoliosis. Echocardiography, performed as a preoperative test, showed aortic valve prolapse and mild aortic valve regurgitation, although heart functions were normal.

Anesthetic course

Although intubation was initially anticipated to be difficult consequent to upper airway thickening and restricted cervical vertebrae mobility, preoperative examinations conducted at the Department of Anesthesiology revealed only mild tongue thickening, with sufficient mouth opening, and diagnostic imaging did not show upper airway thickening (Fig. 2). Anesthesia was induced with 20 mg propofol and 20 μg fentanyl, with manual ventilation possible by mask-to-face ventilation with mandibular elevation only and no posterior neck tilt. Therefore, 2 mg vecuronium was administered for muscle relaxation. The cervical vertebrae were maintained in a good head position and the larynx was gently opened. As glottic thickening was absent and the target area was visible to the naked eye, a 4.5-mm cuffless endotracheal tube was used. For anesthetic maintenance, total intravenous anesthesia was performed using propofol and remifentanil for intraoperative motor-evoked potential monitoring, with no additional muscle relaxant.

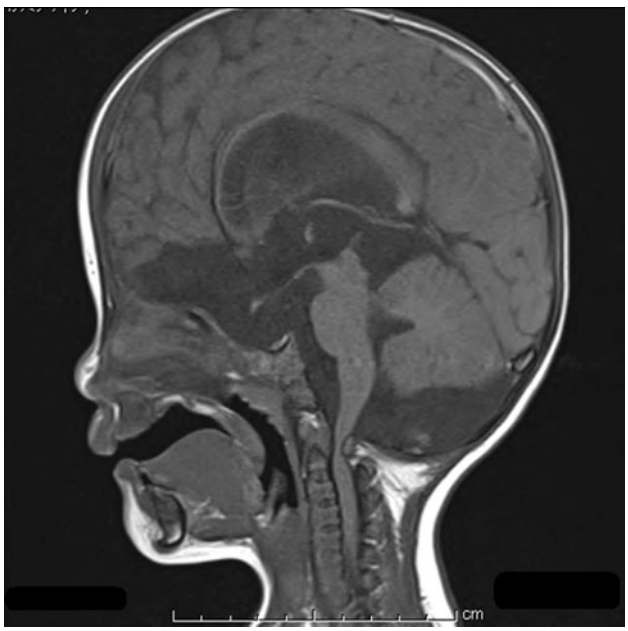

Fig. 1 Magnetic resonance imaging (MRI) scan. Odontoid hypoplasia and ligamentous hypertrophy from clivus to second cervical vertebra (C2) level can be seen, and the spinal cord is severely compressed at first cervical vertebra (C1) level; this was considered the cause of hydrocephalus

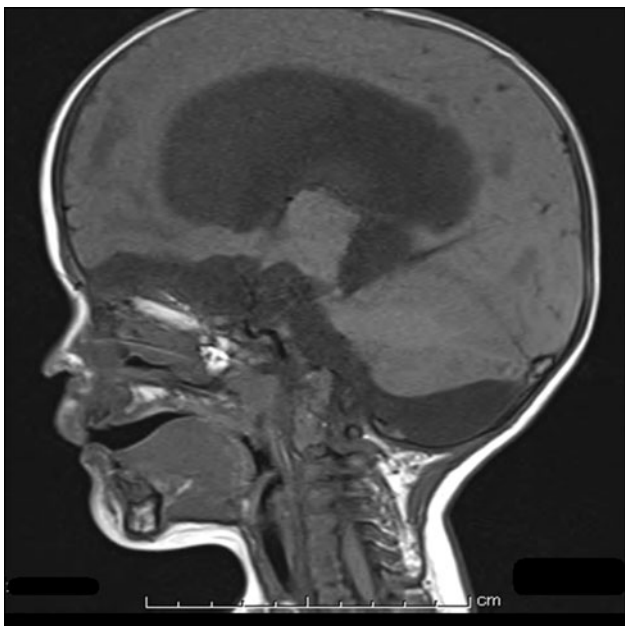

Fig. 2 Magnetic resonance imaging (MRI) scan. Tongue, epiglottis, and vocal cord are not thick, and there is no tracheal obstruction

During surgery, respiratory and circulatory dynamics remained stable.

Sufficient decompression was achieved by a C1–C2 osteotomy, and the surgery was completed without resecting the occipital bone. The operating time was 3 h 9 min and anesthesia time was 5 h 43 min; infusion volume was 552 ml, transfusion volume of packed red blood cells was 60 ml, hemorrhage volume was 80 ml, and urine volume was 277 ml. After surgery, the patient was transferred to the intensive care unit (ICU) while intubated, after which he awoke from anesthesia without difficulty and was extubated 7 h after surgery. The cervical vertebrae were sufficiently protected by keeping the patient at rest in a recumbent position, and postoperative diagnostic imaging showed improvements in cervical compression and motor function of the right upper and lower extremities.

Discussion

Mucopolysaccharidosis is a recessively inherited lysosomal disease, with about 300–400 affected individuals in Japan, for an incidence rate of about 1 in 50,000. Depending on the specific lysosomal enzyme deficiency, mucopolysaccharidosis can be roughly divided into six subtypes, with type VI being extremely rare, accounting for 1% of all forms. In type VI patients, *N*-acetylgalactosamine 4-sulfatase (arylsulfatase B) is missing, causing dermatan sulfate to accumulate throughout the body. A total of six cases of this autosomal recessive inherited disorder had been reported in Japan up to October 2007 according to the Ministry of Health, Labor, and Welfare. The clinical characteristics of mucopolysaccharidosis vary greatly, although they generally include a unique facial appearance, short stature, posterior and lateral curvature of the thoracolumbar vertebrae, upper airway thickening, heart valve disease, hearing loss, and corneal opacity, with specific symptoms varying depending on the site of accumulation. To the best of our knowledge, no mental retardation has been reported with mucopolysaccharidosis type VI. On the other hand, type VI is often associated with spinal damage or hydrocephaly resulting from spinal compression caused by first cervical vertebra hypoplasia, and decompression is required, as in the present patient [1, 2].

The main problem with anesthetic management in individuals with mucopolysaccharidosis is difficulty with airway control, with 25% of reported patients showing a difficult airway and 8% requiring emergency airway control [1, 4]. This problem occurs because most of these patients have airway obstruction and tongue thickening from mucopolysaccharide deposition, along with skull and facial bone deformations, a short neck, difficulty opening the mouth as a result of jaw arthrosclerosis, and anterior displacement of the larynx. As seen with the present patient, tracheal intubation can be complicated by limited neck mobility caused by atlantoaxial joint instability. For airway control, intubation with a bronchofiberscope and awake intubation are recommended [2], although cases in which a laryngeal mask was used for airway control have been reported [4]. In the present patient, who was 9 months old at the time of our procedure, preoperative diagnostic imaging did not show upper airway thickening, and airway control was relatively easy.

As for treatment, in addition to conventional bone marrow transplantation, enzyme replacement therapy using galsulfase has been approved in Japan. Although intubation was easy in the present case, mucopolysaccharides gradually accumulate daily in the body of affected individuals, and airway control may not necessarily remain easy for this patient in the future. Furthermore, it is difficult to evaluate the progression of illness uniformly, as it varies greatly between individuals and is dependent on the period of enzyme replacement therapy. In addition to difficult airway control in mucopolysaccharidosis patients, many display cardiopulmonary dysfunction; thus, thorough testing over time is essential. For selection of anesthetics for use with mucopolysaccharidosis patients, Linstedt et al. [3] recommended anesthesia induction with estazolam, and maintenance using 50% nitrous oxide and 0.4–0.8% isoflurane, with fentanyl for analgesia. In the present study, propofol, remifentanil, and fentanyl were used for anesthetic management, and intraoperative management went smoothly, indicating that currently available clinical anesthetic agents are not problematic.

Conclusion

The present report describes anesthetic management of a patient with mucopolysaccharidosis type VI (Maroteaux–Lamy syndrome). Airway control was unexpectedly easy, and perioperative management based on total intravenous anesthesia using propofol and remifentanil went smoothly without any complications. We also found that preoperative diagnostic imaging was important for effective upper airway assessment.

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