

CASE REPORTS

Anesthetic Management in a Child with Pelizaeus-Merzbacher-like Disease: A Case Report

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Background

Pelizaeus-Merzbacher-like disease (PMLD), a leukodystrophy, is a rare inherited condition primarily affecting the central nervous system. Clinical features such as seizures, hypotonia, spasticity, contractures, respiratory difficulty, dysphagia, and gastroesophageal reflux raise significant anesthetic concerns.

Case

We report the successful management of a 14-year-old child with PMLD scheduled for bilateral foot reconstruction for equinovarus deformity. General anesthesia was induced using rapid sequence induction and maintained with sevoflurane, remifentanyl, and dexmedetomidine infusions. Neuromuscular blockade was not repeated, and the train-of-four count recovered to 4 prior to emergence. Body temperature and fluid balance were closely monitored.

Conclusions

Potential difficult airway, aspiration risk, perioperative seizures, respiratory depression, hypothermia, spasticity, hemodynamic instability, prolonged neuromuscular blockade, positioning challenges, and pain control pose significant challenges to anesthesiologists. Detailed assessment, comprehensive anesthetic planning, and vigilant monitoring are essential for safe anesthetic management in patients with PMLD.

Introduction

Pelizaeus-Merzbacher-like disease (PMLD) is a rare autosomal recessive leukodystrophy characterized by hypomyelination of the central nervous system (CNS). The prevalence is unknown. PMLD shares similar clinical and radiological features with Pelizaeus-Merzbacher disease (PMD) but occurs without proteolipid protein 1 (PLP1) mutations.¹ PMLD is genetically heterogeneous, with reported mutations including those in the connexin 47 gene (GJC2).^{2,3}

Clinical manifestations include global developmental delay, cognitive impairment, seizures, nystagmus, hypotonia with progressive spasticity, contractures, ataxia, dysarthria, dysphagia, and gastroesophageal reflux. Anesthetic considerations include potential difficult airway, aspiration risk,

perioperative seizures, respiratory depression, hypothermia, spasticity, hemodynamic instability, prolonged neuromuscular blockade, positioning challenges, and pain control.

Case Presentation

In July 2022, a 14-year-old Chinese boy with PMLD was scheduled for bilateral foot reconstruction for equinovarus deformity under general anesthesia. He was a dependent, wheelchair-bound resident of a full-time care facility. Since birth, he had progressively developed global developmental delay, feeding difficulties, intellectual disability, and quadriplegia. Metabolic screening results were normal. Brain magnetic resonance imaging (MRI) at 4 years of age showed diffuse hypomyelination without significant cerebral atrophy or focal lesions. Genetic testing revealed no PLP1 mutation.

He required botulinum toxin injections, daytime ankle-foot orthoses, and nighttime knee-ankle-foot orthoses. He had a history of infrequent seizures and had been weaned off anticonvulsants at 11 years of age. Repeated electroencephalograms showed no epileptiform activity. He had frequent drooling and choking, and videofluoroscopic swallow study demonstrated severe dysphagia with silent aspiration. He was managed with a scopolamine patch and nasogastric tube feeding.

On preoperative assessment, his weight was 21.5 kg and height was 134 cm. Baseline heart rate was 120 beats/min, blood pressure was 100/60 mmHg, oxygen saturation was 95% on room air, and body temperature was 36.8°C. He was non-communicative. Laboratory investigations were unremarkable. Chest radiograph showed no consolidation. He was not in respiratory distress. Auscultation revealed mild bilateral airway secretions. He was kept fasting after midnight. The scopolamine patch was continued, and eutectic mixture of local anesthetics (EMLA) cream was applied to the dorsum of both hands.

In the operating theatre, a 20-gauge intravenous catheter was inserted into the left hand. After adequate preoxygenation, general anesthesia was induced using rapid sequence induction with cricoid pressure using fentanyl 20 µg, propofol 50 mg, and rocuronium 35 mg. Tracheal intubation using a video laryngoscope and bougie was uneventful. A size 6.5 cuffed endotracheal tube was secured at 20 cm at the incisors.

Anesthesia was maintained with sevoflurane at 0.7 minimum alveolar concentration (MAC) in 40% oxygen in air. Remifentanyl infusion was titrated between 0.2–0.3 µg/kg/min, and dexmedetomidine infusion was administered at 0.2 µg/kg/h following a loading dose of 0.5 µg/kg over 10 minutes. Bispectral index (BIS) was maintained around 60. Body temperature was monitored using an esophageal probe and maintained between 36.5–37.5°C using active warming devices.

No additional neuromuscular blockade was administered intraoperatively. Intravenous paracetamol 300 mg and morphine 2 mg were given for postoperative analgesia. Levobupivacaine 0.25% (8 mL per foot) was infiltrated by the surgeon at the end of the procedure. Ondansetron 2 mg was administered prior to emergence. Train-of-four monitoring showed recovery to 4 without fade, and no reversal agent was required. Extubation was uneventful.

The duration of anesthesia was 587 minutes due to surgical complexity. Estimated blood loss was 50 mL, and urine output was adequate. In the recovery unit, morphine 1 mg was administered, and oxygen was provided at 3 L/min via nasal cannula. The patient remained stable with oxygen saturation of 95% and no respiratory distress. No adverse events were observed. He was discharged on postoperative day 7.

Discussion

White matter consists of axons wrapped by myelin, which increases the efficiency of neuronal conduction. Leukodystrophies are a group of rare, progressive, degenerative disorders characterized by defective formation of CNS white matter. They differ from demyelinating diseases such as multiple sclerosis. More than 30 leukodystrophies have been described, with symptoms varying depending on the subtype, affected regions, and disease progression.

PMLD is an autosomal recessive leukodystrophy with a phenotype similar to PMD but without PLP1 mutations. Due to its rarity, prevalence remains unknown. It is genetically heterogeneous, with mutations in the connexin 47 gene (GJC2) identified in approximately 8% of cases. A typical MRI finding is diffuse hypomyelination. Nystagmus is often the earliest symptom, followed by ataxia and spasticity.

High aspiration risk is associated with pharyngeal hypotonia, gastroesophageal reflux, and excessive secretions. Premedication with gastric acid suppression may be considered. Oral secretions should be suctioned prior to induction. Rapid sequence induction is preferable. The role of cricoid pressure in pediatric patients remains controversial. Succinylcholine use is debated due to the risk of hyperkalemia in patients with upper motor neuron disease.⁴ Rocuronium provides an alternative with rapid onset and availability of reversal with sugammadex, and sufficient muscle relaxation for intubation can be achieved within about one minute.⁵ Neuromuscular monitoring is essential to ensure full recovery prior to extubation.

Perioperative seizures increase anesthetic risk. A detailed seizure history should be obtained. Anticonvulsants should generally be continued perioperatively. Propofol and barbiturates have anticonvulsant properties. Ketamine, while providing anesthesia and analgesia, may have pro-convulsant and sympathomimetic effects and increase secretions, making it less desirable.

Sevoflurane should be used at low concentrations to minimize CNS excitation, and nitrous oxide is best avoided.^{6,7} Hyperventilation-induced hypocapnia should also be avoided.⁸

Optimizing pain control is challenging. A multimodal, opioid-sparing approach is preferred to reduce respiratory complications. Epidural anesthesia may be effective but was not feasible in this patient due to severe scoliosis. Regional anesthesia is an alternative. Remifentanyl provides rapid-onset, short-acting analgesia, while dexmedetomidine offers sedative and MAC-sparing effects but may have prolonged action. Intravenous or rectal paracetamol may be used. Close postoperative respiratory monitoring is essential following opioid administration.

In literature, there are only a few case reports on the anesthetic management for patients with PMD. There is a case report of a 13-year-old girl with PMD under general anesthesia for resection and recession of ocular muscles in Korea.⁹ Apart from mental developmental delay, there were no other symptoms such as hypotonia, spasticity, seizures, aspiration or feeding difficulty in that patient. In another case report of a 20-year-old gentleman for extraction of impacted teeth under general anesthesia in Japan, the patient had similar symptoms with our patient except no history of aspiration nor gastroesophageal reflux.¹⁰ That patient was co-induced with thiamylal, fentanyl and desflurane. Hyperthermia was reported and linked to exacerbated spasticity due to stress. Although it has been suggested that usual anesthetic drugs are safe for patients with PMD,¹¹ and there is no evidence of PMD-induced malignant hyperthermia, we should remain vigilant with spasticity and hyperthermia. Nonetheless, in both cases the surgeries were short and less painful compared to our case.

Exacerbation of spasticity or dystonia during emergence may result in respiratory compromise or rhabdomyolysis. Benzodiazepines may be used for treatment. Ondansetron is preferred over metoclopramide due to lower risk of extrapyramidal side effects. Active warming is important, as thermoregulation may be impaired and fat insulation reduced. Careful positioning is required to prevent pressure injury. BIS monitoring may be less reliable in leukodystrophy patients.¹²

Conclusion

We presented the anesthetic management of a 14-year-old boy with PMLD. Major anesthetic concerns include potential difficult airway, aspiration risk, perioperative seizure, respiratory depression, hypothermia, spasticity, haemodynamic stability, prolonged neuromuscular blockade, positioning and pain control. Detailed patient assessment, comprehensive anesthetic planning and vigilant monitoring are keys to safe provision of anesthesia.

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Conflicts of interest

The author has disclosed no conflicts of interest.

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Patient consent

Informed consent was obtained for publication of this case report.

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