

Anesthesia for a Patient with Joubert Syndrome Presenting for MRI of a Transplanted Kidney

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Citation

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Abstract

Joubert syndrome is a rare autosomal recessive condition in which there is complete or partial agenesis of the cerebellar vermis. In order to make the diagnosis, there must also be developmental delay, hypotonia, and either abnormal breathing and or abnormal eye movements. (1) The breathing pattern may consist of episodes of both tachypnea and apnea (2). These patients may be sensitive to respiratory depression caused by anesthetics (2). Retinal dystrophy and cystic kidneys may also be associated with this clinical syndrome. This is a report of a patient with Joubert syndrome, presenting for an MRI of a transplanted kidney, who underwent general anesthesia.

CASE REPORT

The patient was a 9-year-old female, who presented for an MRI of a transplanted kidney. She had been previously diagnosed with Joubert syndrome, and exhibited developmental delay as well as ataxia. Although she used a wheelchair, she was able to walk slowly, with assistance. She also had a history of retinitis pigmentosa. She had no previously reported history of breathing abnormalities. Her native kidneys were cystic, and she had developed renal failure, requiring a kidney transplant 2 months prior. Her medications included Prograf, prednisone, Cellcept, bactrim, valganciclovir, pepcid, terazosin, and cefazolin. The patient had developed post-transplant renal failure, and was scheduled for an MRI of the transplanted kidney.

On physical exam, the patient now weighed 34 kg. Her mouth opened wide, with a Mallampati 3 view. The thyromental distance was 5 cm. The neck had full range motion. The cardiopulmonary exam was within normal limits. Lab studies were remarkable for a hemoglobin of 10.1 and hematocrit of 29.2. The sodium was 134, potassium 3.1 mEq/L, BUN 111, and creatinine 2.8.

General anesthesia was necessary for the MRI as there were to be periods lasting 20-30 seconds during which apnea would be required. The patient would not have been able to accomplish this voluntarily. The patient was brought into the MRI room, standard American Society of Anesthesiologists (ASA) monitors were applied. After preoxygenation with a mask, anesthesia was induced with propofol intravenously.

Mask ventilation was performed with oxygen and sevoflurane to further deepen the anesthesia. Tracheal intubation was accomplished via direct laryngoscopy with a Macintosh 2 blade, without the use of muscle relaxation. Anesthesia was maintained with oxygen and sevoflurane. Ventilation was mechanically controlled, and one additional bolus of propofol was administered prior to the first breath-holding request. The patient remained apneic during the requests for breathholding. At the conclusion of the scan, which lasted 36 minutes, the sevoflurane was turned off. The awakening of the patient appeared delayed, and the trachea was extubated 28 minutes later. The scan did not reveal any abnormalities that could account for the renal failure. The patient recovered uneventfully from the anesthetic.

DISCUSSION

The first report of patients with this clinical syndrome was by Marie Joubert in 1969 – a report on four siblings of consanguineous parents (3). This syndrome was subsequently termed Joubert syndrome in 1977 by Bolthausen and Isler (4). It is a rare autosomal recessive disorder, occurring more frequently in the children of consanguineous parents. Its onset is in the neonatal period and has a poor prognosis (2,3). Patients with Joubert syndrome have varying degrees of cerebellar vermis dysplasia. Clinically, patients exhibit developmental delay, hypotonia, ataxia, irregular breathing patterns, abnormal ocular movements, retinal dystrophy, and cystic kidneys. Our patient displayed many of these features, although there was no reported history of breathing

abnormalities. The possibility of irregular breathing and hypotonia may be exacerbated by opioids and muscle relaxants.

Although first reported 37 years ago, there have been only four previous anesthetics described of patients with Joubert syndrome (2,5,6). The first report was of an infant who underwent general anesthesia for repair of an inguinal hernia (3). In that case, no premedication or narcotics were administered. An ilio-inguinal /iliohypogastric nerve block was done prior to the surgery. Anesthesia was induced with thiopentone, and maintained with nitrous oxide, oxygen, and isoflurane. Postoperatively, there were many apneic episodes which persisted for 'some hours', but then lessened. In 1997, Habre et al (2) reported two children with Joubert syndrome who had abnormal respiratory patterns after general anesthesia. Vodopich et al (6) performed a spinal anesthetic on a 7 month old infant. Although no intravenous sedation was administered, there were brief episodes of apnea. These authors recommended considering the use of caffeine, to reduce the apneic periods. Our case required general anesthesia with breath-holding sequences. The main concerns in our case included possible unusual airway anatomy and abnormal respiratory patterns.

Joubert syndrome has been associated with various airway abnormalities – high, arched palate, large or protruding tongues, laryngomalacia, and micrognathia (3,7). These abnormalities may cause difficulty with tracheal intubation. We did not administer muscle relaxants, to minimize the chances of postoperative weakness. One of the most important issues to consider is the abnormal breathing pattern of patients with Joubert syndrome. In most cases, the breathing abnormality improves with age. The apneic episodes may be prolonged by administering opioids (3). We were able to avoid the use of opioids but the procedure still required general anesthesia. We administered sevoflurane alone with oxygen for maintenance of anesthesia, and propofol boluses for the required breath-holding sequences.

After turning off the anesthetic, there appeared to be prolongation to time of awakening and therefore extubation, recorded at 28 minutes. Standard post-anesthesia monitoring in the recovery room demonstrated no complications and the patient was discharged to the floor after an uneventful recovery period.

CONCLUSION

Joubert syndrome is a rare disorder with early onset and poor prognosis. Associated with the syndrome are specific concerns for the anesthesiologist. These include the possibility of unusual airway anatomy, hypotonia, and abnormal respiratory patterns which may be exacerbated by general anesthesia. It may be prudent to avoid the use of opioids and muscle relaxants – to decrease the risk of post-anesthesia hypotonia and abnormal respirations.

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