Joubert syndrome (JS) is a rare autosomal recessive condition that is caused by dysplasia of the cerebellar vermis with cystic malformation of the brainstem giving rise to the typical molar tooth sign on the MRI. Clinical features include hypotonia, ataxia, severe mental retardation, abnormal eye movements, episodic tachypnea alternating with apnea, multicystic renal disease and hepatic fibrosis. Patients with JS may require anesthesia or sedation for surgery or for investigative radiological procedures.

We compare the anesthetic management of a patient with JS for dental procedures that were two years apart and discuss the differing outcomes, emphasizing the anesthetic considerations and the challenges that these patients pose to the anesthesiologist.

The patient had clinical (hypotonia, apnea) and radiological (deformity of the fourth ventricle with hypoplasia of the cerebellar vermis) signs of JS at birth. At the age of 19 months, he developed lower lip trauma secondary to biting down with his maxillary incisors. He presented for full mouth restoration and extraction of his primary anterior maxillary teeth under general anesthesia. During the anesthetic he received 1 mg morphine IV and local infiltration with 1% lidocaine for analgesia. He was extubated successfully. In the recovery room, he received 5mg ketorolac IV and 120 mg acetaminophen PO for pain. While in the recovery room, he developed apnea and bradycardia with desaturation (SpO2 of 75%). He was successfully resuscitated by bag mask ventilation with 100% oxygen. He continued to maintain his oxygenation and was later discharged home.

At the age of 4 years, he presented again, with lower lip ulceration and scarring from biting down on his lower lip with his maxillary canines. The maxillary canines were extracted and full mouth restoration was performed under general anesthesia. Local infiltration with 1% lidocaine was used for analgesia, IV narcotics were avoided. The anesthetic was uneventful. He received 200 mg acetaminophen PO for pain in the recovery room. There was no episode of apnea in the postoperative period.

The brainstem abnormalities in patients with JS result in abnormal respiratory patterns. These patients are particularly sensitive to anesthetic agents and opioids, increasing the chances of post-operative apneic episodes. The morphine administered during the first anesthetic could have contributed to his episode of apnea and hypoxia. However, patients with JS have also been shown to have decreasing episodes of apnea with age. The patients second dental anesthetic was more than 2 years later when he was 4 years old.

References: