Anesthetic Management in Joubert Syndrome: Case Report

Joubert Sendromunda Anestezi

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ABSTRACT Joubert syndrome is a rare autosomal recessive disorder of cerebellum that occurs in 1 of 100.000 live births. The syndrome is characterized by a distinctive cerebellar and brainstem malformation, hypotonic, developmental delays, either hyperpnea or apnea or atypical eye movements or both. Cognitive abilities are variable, ranging from severe to mild mental retardation. A magnetic resonance imaging (MRI) is the most common neuroimaging technique used to establish the diagnosis of Joubert syndrome. "Molar teeth" sign on MRI is spesific for Joubert Syndrome. There are limited reports on the anesthetic manegement of these patients. These patients may be sensitive to respiratory depression caused by anesthetics. Because of these reasons, anesthetic management of these patients need more attention. These patients are especially susceptible to N_2O, opioids and neuromuscular blockers. Ventilation parameters must be monitored closely for at least 24 hours postoperatively.

Key Words: Abnormalities; cerebellum; anesthesia, general; analgesics, opioid

ÖZET Joubert sendromu serebellumun otozomal resesif hastalığı olup 100.000 doğumda bir görülür. Sendrom serebellum ve beyin sapının malformasyonu, hipotoni, gelişimsel gecikme, hiperpne veya apne atakları veya atipik göz hareketleri ile karakterizedir. Kognitif değişiklikler hafiften şiddetli zeka geriliğine kadar değişkenlik gösterebilir. Joubert Sendromunun tanısı için en sık kullanılan nörogörüntüleme yöntemi manyetik rezonans görüntüleme (MRG) dir. MRG'de molar diş işareti Joubert sendromu için spesifiktir. Literatürde bu hastaların anestezi yaklaşımı ile ilgili sınırlı sayıda yayın bulunmaktadır. Bu hastalar anestetiklerle oluşan respiratuar depresyona duyarlı olabilirler. Bu sebeple bu hastaların anestezi idamesi dikkatli takip gerektirir. Bu hastalar özellikle N_2O, opiodlere ve kas gevşeticilere duyarlıdırlar. Postoperatif en az 24 saat ventilasyon parametrelerinin takibi önerilmektedir.

Anahtar Kelimeler: Anomali; serebellum; anestezi,genel; opiod analjezik

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Joubert syndrome is a rare autosomal recessive disorder of cerebellum that occurs in 1 of 100.000 live births.[1] NPHP1, CEP290, AH11, TMEM67 genes are accused for the syndrome.[2,3] The syndrome is characterized by a distinctive cerebellar and brainstem malformation, hypotonic, developmental delays, either hyperpnea or apnea or atypical eye movements or both. Cognitive abilities are variable, ranging from severe to mild mental retardation.[2,4,5] These patients may be sensitive to respiratory depression caused by anesthetics[6] and because of their anatomic feature (lar-

ge protruding tongue, laryngomalacia, micrognathia) airway management may be difficult. In our report, the anesthetic management of a Joubert syndrome who underwent a surgery of ingunal hernia, hydrocele and circumcison was presented.

CASE REPORT

A 7-year-old boy who suffered from inguinal hernia and hydrocele admitted to our hospital. The urologist planned his circumcision at the same time. He was born at a gestation of 38 weeks by Ceasarean section from the first pregnancy of his mother. His mother's pregnancy was normal and there was no consanguinity between the parents. He was first admitted to paediatric clinic with developmental delay when he was 6 months old. During his examination, cranial magnetic resonance imaging (MRI) was obtained cerebellar vermian hypoplasia was reported on cranial MRI, the inferior part of the vermis was incomplete and cerebellar hemispheres showed opposition. Molar teeth sign was seen in the 4th ventricle which was significant for Joubert syndrome. He had a special education, he was able to cooperate. He had another surgery at the age of 4 due to inguinal hernia. As learned from the anesthesia report, sevoflurane and fentanyl was used for induction and no complications were reported in the postoperative period.

On his physical examination, he had hypertrophic tonsils, hypertelorism, high arched palate and a large tongue, and his Mallampati score was class I. After in formed consent from was obtained from his family he was taken to the premedication room and IV access was maintained easily with perfect cooperation. After premedicated with 0.07 mg/kg midazolam IV he was taken into the operating room. After the patient was monitored, propofol 3 mg/kg and sevoflurane was used for both induction and for maintenance at 1.5 MAC (minimum alveolar concentration). During direct laryngoscopy epiglottis was easily seen and graded as I. No 2.5 no laryngeal mask was applied. Opiods were avioded both during induction and for maintenance. Sevoflurane at 1.5 MAC in O_2 and air without N_2O were used. Propofol boluses were needed during surgery. The patient also recieved penile block for analgesia. The surgery lasted for 90 minutes. The patient was allowed to breath spontenously during the surgery and no desaturations were recorded. As the surgery ended, laryngeal mask was removed. After 10 minutes, he was able to respond to verbal commands. He was transferred to postanesthesia care unit (PACU) with spontaneous breathing with a saturation of 96% at room air. He was closely monitorized for ventilation parameters including end tidal carbon dioxide ($ETCO_2$), number of breathing per minute and pulse oximeter. There was no apneic or hyperpneic periods during his follow up at PACU. After one night stay at PACU, he was tranfered to ward without any complications. He was discharged from hospital on 36th hour of surgery.

DISCUSSION

Joubert syndrome was described by Marie Joubert in 1969. It was described as an autosomal recessive disorder that included physchomotor developmental delay, hypotonia, oculomotor abnormalities, retinal distrophy and cystic kidneys. These patients have abnormalities of respiratory control resulting in episodic tachypnea and apnea, particularly in neonatal period.[1,4,5] Patients with Joubert Syndrome have varying degrees of cerebellar vermis dysplasia. MRI is the most common neuroimaging technique used to establish the diagnosis of Joubert syndrome.[7,8] "Molar teeth" sign on MRI is spesific for the disease.

There is not much references about anesthetic management in Joubert syndrome. The first report was published in 1989, and it was about inguinal hernia repair in an infant.[9] General anesthesia was maintained with thiopentone, N_2O, O_2, isoflurane and papaveretum, and there were many apneic episodes persisted for hours in this patient.[9]

In 1997, Habre and friends reported two patients wih Joubert syndrome. One child had several apnea and hyperpnea episodes postoperatively in whom opioids had been used for induction and postoperative analgesia.[6] There are some conflicting reports about using inhalation agents in these cases. Habre and friends reported that inhalational induction resulted in apnea, so they had to use that

IV induction, but they still suggested inhalational induction because of faster recovery.[6]

In 2004, regional anesthesia was maintained for inguinal hernia repair by Vodopich and friends.[10] They recommended spinal anesthesia under propofol sedation with spontaneous ventilation. They have met several apneic episodes during surgery but no desaturation below 95 was observed.[10]

Another report for anesthetic management in Joubert syndrome was about anesthesia for MRI. Jeng and friends mentioned IV propofol for induction and sevoflurane for maintenance.[10] They reported delayed extubation for 28 min and they concluded that opioids must be avoided, similar to other reports.[11]

Karamehmet and friends reported that they used N_2O for three times repeatedly for a baby with Joubert syndrome, and they did not encounter any respiratory depression.[12]

As Joubert syndrome has been associated with various airway abnormalities like high arched palate, large protruding tongue, laryngomalacia, micrognathia[1,3,4] which may lead to difficult airway, we were prepared for difficult intubation.

We planned to keep and asist spotaneous ventilation without using any opioids because these patients are frequently subject to apneic episodes.[6,9,10] We also preferred not to use N_2O and performed penile block under sedation. Anesthesia was maintained by sevoflurane at 1.5 MAC in air and with propofol bolus doses when needed. Short term apneic episodes were seen after propofol bolus doses and was assited by manual ventilation all through the operation, mostly in order to maintain spontaneous ventilation. We did not allow any desatırations in these episodes.

As surgery ended, he was allowed to breath spontaneously. Laryngeal mask was easily removed and he was able to respond quickly to verbal commands. Besides penile block we also used intramuscular diclofenac sodium 3 mg/kg for postoperative analgesia. In PACU no apneas or hyperpneas were observed.

As a result, during anesthetic management of Joubert Syndrome we recommend IV induction without using opioids or neuromuscular blocking agents and maintaining spontaneous ventilation. As for maintenance of anesthesia, inhalation agents without N_2O are preferred combining with regional or local blocks according to surgery. Ventilation parameters must be closely monitored for at least 24 hours postoperatively.

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