

Anaesthesia Management in Patient of Joubert Syndrome Posted For Continuous Ambulatory Peritoneal Dialysis (CAPD) Catheter Insertion

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Abstract

Joubert syndrome (JS) is a rare autosomal recessive genetic disorder which mainly includes cerebellum. In Joubert syndrome partial or complete agenesis of the cerebellar vermis occurs. In this syndrome there occurs malformations of the cerebellum and brain stem. This syndrome is characterized by hypotonia, ataxia, mental retardation, abnormal eye movements and a respiratory pattern of alternating hyperpnea-apnea. These patients may be sensitive to respiratory depression caused by anaesthetics agents, so the anaesthetic management in these patients are challenging and needs more attention. In this report we are presenting the case of 8 yr. old male child with Joubert syndrome posted for CAPD(continuous ambulatory peritoneal dialysis) catheter insertion surgery under General Anaesthesia.

Keywords: Joubert syndrome, anaesthesia management, CAPD surgery

Introduction

Joubert syndrome (JS) is an inherited autosomal recessive disorder .It is disorder of brain development that may affect many parts of the body. It is characterized by the underdevelopment or absence of the cerebellar vermis and malformed brain stem ^[1]. Signs and symptoms can vary but commonly includes hypotonia, abnormal breathing patterns, abnormal eye movements, ataxia, distinctive facial features, developmental delay and intellectual disability ^{[2][3][4]}. Magnetic Resonance Imaging (MRI) of the Brain in the patients with Joubert syndrome shows the "molar tooth sign" and "bat wing" appearance of fourth ventricle on coronal and axial cranial sections ^[5]. These patients may be susceptible to respiratory depression with volatile and intravenous anaesthetic agents^{[6][7]}. Regional anaesthesia is strongly recommended in these patients, in combination with sedation, because of the psychological disturbances of the patients. We report a

case of 8 yr old male child with Joubert Syndrome who underwent CAPD catheter insertion surgery under General Anaesthesia.

Case Report

A 8 year old male child, 19.5 kg with Joubert Syndrome admitted in Urology department in Santokba Durlabhji Memorial Hospital, Jaipur for Percutaneous Continuous Ambulatory Peritoneal Dialysis (CAPD) catheter insertion surgery. The child had been delivered by caesarean section at 40 weeks gestation. The pregnancy had been of normal course and consanguinity was not there between the parents. The patient was hypotonic at birth and stayed in the Intensive Care Unit for 10 days because of respiratory disorders at birth. Brain and renal pathologies were detected prenatally and Joubert Syndrome was diagnosed with cranial magnetic resonance imaging (MRI) at the age of 8 months. The physical examination revealed hypertelorism, large tongue, high arched palate, delayed milestone, bilateral squint and ataxia.

Patient had history of 2 seizure episodes in past 2 month for which he was on antiepileptic medications. There was no past history of anaesthesia exposure other than sedation with inj. midazolam (0.04 mg/kg) during MRI. The Mallampati score was class 1 and laboratory test results were normal. As per the standard institutional protocol during pre-anaesthetic checkup the patient was ordered fasting. On the day of surgery, morning dose of antiepileptic medication was given, written informed consent was taken from patient parents and then patient shifted to operating room. Standard monitors were attached and baseline data were taken, blood pressure, heart rate and SpO₂ were 110/78 mmHg, 106 beats/min. regular and 97% respectively.

Intravenous cannula of 22 gauge was inserted and premedicated with inj. Atropin (0.02 mg/kg), inj. midazolam (0.04 mg/kg), inj. Ondansetron (0.1 mg/kg), inj. fentanyl (1 µg/kg). Patient induced with inj. Propofol 30 mg, inj. Atracurium 8 mg (0.4 mg/kg) and intubated with uncuffed endotracheal tube no. 5.5 under vision. Anaesthesia was maintained with 1 MAC sevoflurane in 50% oxygen- 50% air mixture. The surgical procedure lasted for 40 min. After spontaneous breathing of the patient, reversal with inj. neostigmine 1 mg and inj. glycopyrrolate 0.16 mg was given and patient extubated smoothly. Postoperatively patient was conscious, awake and hemodynamically stable. Postoperative analgesia of 200 mg (10 mg/kg) Paracetamol in 100 ml normal saline was administered and the patient was transferred to the post- anaesthesia care unit (PACU) for observation. When the patient was considered stable, he was transferred to the Urology ward and discharged from the hospital 48 hours postoperatively.

Discussion

Joubert syndrome was originally described in 1968 in siblings with agenesis of the cerebellar vermis, presenting with abnormal breathing patterns, abnormal eye movements, ataxia, and intellectual disability^[8]. The abnormal breathing pattern may cause problems during the post-operative period. Patients with Joubert syndrome may be susceptible to respiratory depression with anaesthetic agents (especially opioids and neuromuscular blocking drugs)^{[9],[10]}.

Respiratory depression and the central apnea seen in these patients may be prolonged by the administration of long acting opioids.

A marked sensitivity to opioids in Joubert syndrome patients was described in some case reports as manifesting mainly as apneic episodes^[10]. Fentanyl is a

relatively short acting synthetic opioid, its action wears off early, thus decreasing the risk of potentiating apnea in patients with Joubert syndrome. It has been emphasized that muscle relaxants and opioids should be avoided in the anaesthesia management of patient with this syndrome and if at all they are used then short acting drugs should be preferred.

In above case we used Fentanyl and Atracurium because of their safety profile and short duration of action. Our patient had no apnea episode in the postoperative period and was quite comfortable. Our patient received Paracetamol to supplement analgesia in postoperative period, as there was no hepatic involvement as evidenced by blood tests and ultrasound of the abdomen.

Other clinical features of Joubert syndrome that may be relevant to the conduct of anaesthesia include palate malformations, a large or protruding tongue, laryngomalacia, and micrognathia. In spite of our patient's high arched palate and large protruding tongue, we did not face any difficulty with airway management. The Mallampati score of our patient was class 1 and as mask ventilation was easy, there was no difficulty in intubation.

Conclusion

Joubert syndrome is a rare autosomal recessive disorder with multisystem involvement. These patients may be susceptible to respiratory depression with General anaesthesia drugs therefore Regional anaesthesia is strongly recommended in these patient as much as possible. Short acting drugs should be preferred. The abnormal breathing pattern with central apnea renders anaesthetic management complicated in the perioperative period. The short acting profile of Fentanyl is useful in situations when opioids are deemed necessary. Consideration of NSAIDs as a part

of multimodal analgesia, when renal and hepatic systems are not involved, may be of benefit. Other important group of drug which can helpful in these patients is alpha-2 adrenergic agonist like Dexmedetomidine and Clonidine. They does not affect respiratory drive and has an opioid-sparing effect.

A careful pre-anaesthetic evaluation, a competent understanding of the possible adverse effects of the disease, proper preparedness and vigilance will reduce peri-operative problems and will help in successful patient outcome.

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