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Anaesthesia for a Patient With Jouberts Syndrome Presenting for Cleft Palate Repair-A Case Report

KEYWORDS: Jouberts syndrome / Molar sign/ Cleft palate repair/ Ataxia

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ABSTRACT *Jouberts 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. Retinal dystrophy and cystic kidneys (1) may also be associated with this clinical syndrome. Here we present a case report of a patient with Joubert syndrome, for Cleft palate repair, who underwent general anaesthesia.*

Introduction

Jouberts Syndrome is characterized by cerebellar agenesis (molar tooth sign on MRI), ataxia, developmental delay, oculomotor apraxia, large head with upturned nose and evident nostrils, tongue protrusions with rhythmic movements, pigmentary retinopathy. Airway management in this setting is a challenge to the anaesthesiologist because of laryngomalacia and other anatomical developmental abnormalities; Intraoperatively abnormal vagomimetic action can result in severe bradycardia and post operatively leads to apneic spells and delayed recovery. Hence, we present a case report of a patient with Joubert syndrome, admitted and posted for Cleft palate repair, who underwent general anesthesia.

Case Report

This 3-years-old baby girl weighing 10 kgs, height of 90 cm was admitted into Department of plastic surgery, Owaisi hospital and research centre, Hyderabad for the repair of cleft palate. As per the history taken, patient was born to non-consanguineous parents. At birth the patient was reported to have had difficulty in breathing and swallowing. It was reported, at that time the Neonatologist and ENT surgeons opined that the child had laryngeal hypoplasia. As the child was not expected to survive, hence no therapy was suggested.

On examination the baby was found to have large prominent head and protruding forehead, high-rounded eyebrows, epicanthic folds, bifid tongue, Cleft palate, delayed milestones, strabismus, not holding her head steady, able to sit up and polydactyly (seven digits each in both hands and both feet) but able to hold objects with a pinch, difficulty in breathing and swallowing.

On the day of surgery patient was pre-medicated with Atropine IM (0.2 mg) 20 minutes before the surgery, Pre-oxygenation was done for 3 minutes. Patient was induced with Sodium thiopentone (50mg) IV followed by Suxamethonium (10mg) IV. The patient was intubated with RAE ET tube No: 3.5 uncuffed, throat pack was done and then connected to the Jackson-Rees modification of Ayre's T-Piece with a flow of N_2O (50%), Oxygen (50%) and Sevoflurane (2-3%). A smaller dose of Atracurium (2.5 mg) was given IV.

But, when the Dingman mouth gag was applied with the neck in extension, the patient went into bradycardia which was persisting to less than 30 beats. Hence, mouth gag was removed, oxygenation was continued and the patient was given injection Atropine (0.3mg) IV, patient heart rate improved to 130 per minute. After sometime when the heart rate was maintained within the range of 100-130, the mouth-gag was reapplied and the patient again went into bradycardia, which was persisting and the same procedure was followed by giving Atropine (0.3 mg) and the heart rate increased to 100-130 per minute. At this time the surgery was deferred. IPPV was continued. The patient was reversed by Atropine and neostigmine IV. The patient took more than 30 minutes than normal time for recovery. After the patient was completely recovered she was shifted to the recovery room and monitored with standard recommended techniques and closely watched for apneic spells.

After detailed discussion with the surgeon, patient was advised to come back after 12 months. This period was chosen to appreciate growth and development.

After 14 months case was taken up again with a slight change in anaesthesia plan. Patient was kept NPO as regular. Patients weight was 11kgs, premedication was given with Atropine IM (0.2mg) 20 minutes prior to surgery. Pre-oxygenation done for 3 minutes. Patient was induced with inhalational technique using Sevoflurane 4-6% only. After patient became deep Intubation was successfully done with RAE ET 4.5 mm ID and connected to anaesthesia machine using pediatric circuit. Maintenance was with N2O (50%), Oxygen (50%) and Sevoflurane 2-3%. Neither narcotics nor muscle relaxants were used. After stabilizing and

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positioning the patient, surgeon took over, same gag was applied with no complications. Duration of surgery lasted for 45 minutes. Patient was haemodynamically stable intraoperatively. Surgery was successfully done and patient was extubated after full recovery. Postoperatively patient was satisfactory and no apnoeic spells and hypotonia were noticed. Patient got discharged after 3 days and is being followed up regularly. She might be posted for other procedures in future.

Discussion

The first report of patients with this clinical syndrome was reported by Marie Joubert in 1969 – a report on four siblings of consanguineous parents (2). This syndrome was subsequently termed as Joubert syndrome in 1977 by Bolthauser and Isler (3). 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 (2,4). 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, initially, only four previous cases of Joubert syndrome have been described by the anesthetists (2, 5, 6, 7). Another 12 cases had been reported in 2001 [8], which was a retrospective study correlating clinical features and the neuroradiological (MRI) findings. 7 new cases were reported in Turkey in 2004 (9). The first report was of an infant who underwent general anesthesia for repair of an inguinal hernia (5). In that case, neither premedication nor narcotics were administered. An ilio-inguinal or 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 Jouberts syndrome who had abnormal respiratory patterns after general anesthesia. Vodopich et al (6) performed spinal anesthesia 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 for cleft palate repair, hence prior to the surgery an MRI was done to rule out any airway and cerebellar abnormalities, but because of the two episodes of bradycardia secondary to vagal stimulation, surgery was deferred to a later date. No data is available regarding the cardiac complications of Jouberts syndrome. The main concerns in our case included recurrent bradycardia. It was decided to postpone the procedure in view of the above problem. Considering the fact that the patient has had progressively improved control on respirations over the last three years, it is possible that we may be able to proceed with the surgery after another 6 to 12 months.

Joubert syndrome has been associated with various airway abnormalities – high arched palate, large or protruding tongue, laryngomalacia, and micrognathia (3, 7). These abnormalities may cause difficulty in tracheal intubation. 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, if opioids are administered (5). The procedure required general anesthesia. Hence, we administered Sevoflurane with oxygen and nitrous oxide for maintenance of anesthesia, and a small dose of atracurium for adequate relaxation. Once it was decided to defer the surgery and after turning off the volatile anesthetic, the patient had delayed recovery (30 minutes) and prolonged apnea. Thereafter the patient was reversed and extubated. Standard post-anesthesia monitoring in the recovery room demonstrated no complications and the patient was discharged to the post operative ward for subsequent management.

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. But the unusual observation in this case was recurrent bradycardia, responsive to parasympatholytic agent. It may be prudent to premedicate with vagolytics and avoid the use of opioids and regular dose of muscle relaxants – to decrease the risk of post-Operative anesthesia hypotonia and abnormal respirations.

Fig. 1 Photograph of the patient showing cleft palate, high arched palate, overcrowding of the teeth, upturned nose with prominent nostrils.

Fig. 2 Photograph of the patient showing cleft palate,

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high arched palate, overcrowding of the teeth, ocular apraxia bifid tongue, Nystagmus and Staring look.

Fig. 3a: Polydactyly of the Bi-lateral and feet (classically 7 digits of all four limbs).

Fig. 3b: Polydactlyly of the Bi-lateral Hands (left hand shown in picture)

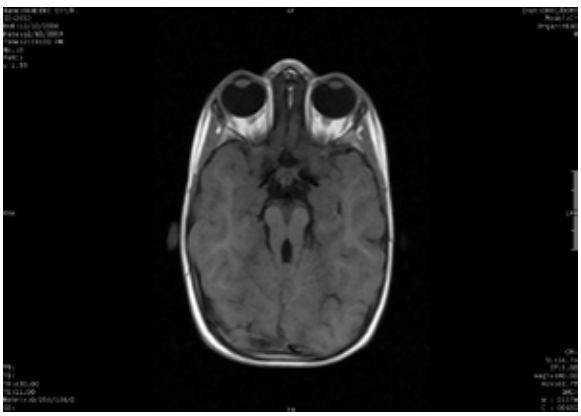

Fig. 4 MRI of the patient showing "Molar Tooth" Appearance on axial images.

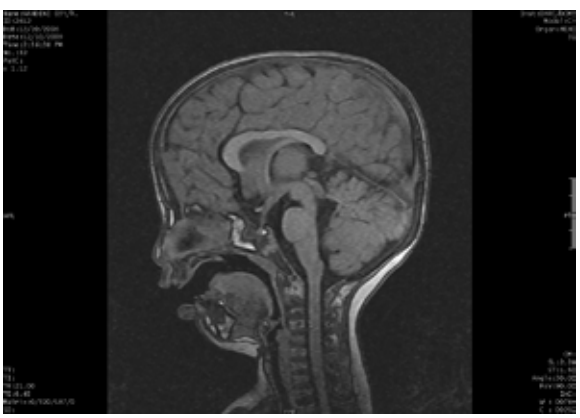

Fig. 5 Sagittal view MRI showing high arched palate, Large tongue, subglottic narrowing

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