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Anaesthetic Management of a patient with Rubinstein Taybi Syndrome

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Abstract

Presentation
We present a case of an 11 year old child with Rubinstein Taybi Syndrome that presented to us for Manipulation under Anaesthesia of fractured Radius and Ulna.

Diagnosis
Diagnosed case of Rubinstein Taybi Syndrome during the first year of life. She was evaluated by paediatric cardiologist during childhood and was cleared of any cardiac abnormalities. There was no murmur heard on auscultation.

Treatment
Perioperative Anaesthetic Management of a patient with Rubinstein Taybi Syndrome under General Anaesthesia.

Discussion
Safe Perioperative Anaesthetic management of a patient with Rubinstein Taybi syndrome which is a rare genetic disorder with challenging anaesthetic considerations.

Introduction

Rubinstein-Taybi syndrome (RTS) is a rare genetic disorder. [1,2] The clinical features of syndrome include growth delay and intellectual disabilities. It also presents with multiple anomalies such as microcephaly, micrognathia, hearing disabilities, congenital heart problems.[3]

There is limited literature on the anaesthetic management of RTS patients due to it being a rare disorder [4, 5,6]. Airway management is challenging as they are at a higher risk of obstructive sleep apnoea due to cranio-facial abnormalities which include micrognathia and macrostomia.[10]

Patients with RTS also have a higher prevalence of congenital heart disease as well as conduction abnormalities.[11] They also have intellectual disability along with speech and hearing disability. Furthermore, they also exhibit behavioural issues such as noise intolerance, short attention span and moodiness. This makes cooperation with anaesthetic induction challenging and they may require premedication and a well formulated plan of anaesthesia. A preoperative visit and discussion with the parents/ guardian are of utmost importance. [12]

Case Report

An 11-year-old, 35 kg weight , 1.2 meter tall girl presented to us with a history of fall 2 days back and had a right radius and ulnar fracture. She was diagnosed with Rubinstein Taybi syndrome at the age of 1 year. She had severe intellectual disability. Examination revealed microcephaly, micrognathia, macroglossia and narrow hard palate. Mouth opening was adequate. Her Mallampatti was 2 and there was no limitation in neck flexion and extension. Biochemical labs were within normal limits. She was evaluated by paediatric cardiologist during childhood and was cleared of any cardiac abnormalities. On auscultation there was no abnormal heart sound.

Keeping in consideration of a difficult airway, difficult airway trolley was kept in the theatre with all sizes of equipment available. Written informed consent was taken from mother.

10 mg Per Oral Midazolam (0.3mg per kg) in ward. AAGBI monitoring was attached in theatre. Inhalational Induction with Sevoflurane 4% was done. 22G cannula was inserted on the dorsum of left hand and secured. Bag mask ventilation was easy. Igel size 2.5 was used to secure the airway. Anaesthesia was maintained with sevoflurane 2.5%. The surgeons performed manipulation of the right radius and ulna and the procedure lasted for 40 minutes. Patient remained hemodynamically stable. Ondansetron 3.5mg IV, dexamethasone 3.5 mg IV , Diclofenac 37.5 mg PR and paracetamol 680 mg PR were given. At the end of the procedure patient was shifted to recovery with igel and igel was removed when patient had spontaneous eye opening. She had a smooth uneventful recovery and was discharged home the same day with regular paracetamol and ibuprofen for pain control.

Discussion

Prevalence of Rubinstein–Taybi syndrome is as rare as 1/250,000–300,000. [13] The Anaesthetic implications include airway challenges, risk of aspiration and cardiac defects along with risk of arrythmias with anaesthetic medications.[14]

As our patient was uncooperative and had no history of Gastroesophageal disease (GERD) we chose premedication followed by inhalational induction and maintenance of spontaneous ventilation until IV line was secured.

There are multiple reports of difficult bag mask ventilation and laryngoscopy in patients with RTS. This is due to limited mouth opening, hypoplastic mandible and high arched palate.[15] Although literature suggests Endotracheal intubation superior to a laryngeal mask airway or Igel due to patients being at high risk of GERD, we used Igel as our patient had no history of GERD.[15]

In 24-38% of patients, cardiac defects have been reported which includes structural as well as conduction abnormalities.[16] In such patients, use of anti-cholinesterase and anticholinergic drugs increases the risk of arrythmias.[14] No preoperative and postoperative problems were encountered in our patient.

Rubinstein-Taybi syndrome is a rare genetic disorder with significant anesthetic considerations. These include a thorough preoperative evaluation, with particular attention to a history of GERD, cardiac abnormalities, and the need for a cardiac consultation. An airway assessment should be conducted, and a management plan for airway support should be formulated. Premedication and involvement of parents or guardians in the anesthesia plan are also essential. The procedure should be performed in a facility experienced in managing difficult airways. Medications such as atropine, neostigmine, and succinylcholine should be avoided.

Declarations of Conflicts of Interest:

None declared.

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