Apert's Syndrome and Anaesthetic Implications

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Abstract

Apert's syndrome is an autosomal dominant genetic disorder characterized by midface hypoplasia, exophthalmia, hypertelorism,choanal atresia and symmetric syndactyly of the hands and feet.Airway obstruction is frequently observed (40%)in patients with severe craniosynostosis syndromes like Apert's and Crouzon. It is important for health care providers treating these patients to recognize the risk factors and potential complications before sedation or anesthesia as repeated surgical corrective procedures are commonly undertaken in these patients. Here, we report an interesting case of Apert's syndrome, posted for an ectropion correction surgery.

Keywords : Apert's syndrome- airway management, anaesthesia

Introduction

Apert's syndrome or acrocephalosyndactyly type 1, first described in 1906, is a rare syndrome manifested with severe craniosynostosis, due to mutation of fibroblastic growth factor type-2 gene. It is a hereditary defect in the tissues that separate each of the various bone analages before the fifth and upto the sixth week of embryonic life. The incidence is estimated to be 1:160000 births, and no predilection by gender has been observed [1].

Case report

A 20 year old male, diagnosed with Apert's syndrome was posted for ectropion correction of right eyelid.Pre-operative evaluation revealed an alert and active male weighing 50 Kg.The patient hadmacroglossia, craniosynostosis manifested with peaked vertically elongated head, short wide nose with bulbous tip, ocular proptosis,wide set eyes, maxillary hypoplasia, mandibular retraction with nasal intonation to his voice (Figure 1).He also had symmetric syndactyly of the hands and feet (Figure 2) and chest deformity (pectusexcavatum). Restricted mouth opening of 2 cm was noted along with deviated nasal septum. Intraoral examination showed high arched palate (Figure 3). However, his neck movements and mentohyoid distance was normal. He also had a history of sleep apnoea and preferred to sleep in lateral position, however, he had no symptoms related to respiratory tract infections. He had undergone two surgical corrective procedures 4 years back for proptosis and dental mal-alignment, under general anaesthesia. The biochemical laboratory parameters including arterial blood gases, ECG and 2D ECHO cardiogram were essentially normal. Neck X- ray revealed fused C4-C5 vertebrae.

Figure 1. Midface hypoplasia, proptosis and hypertelorism

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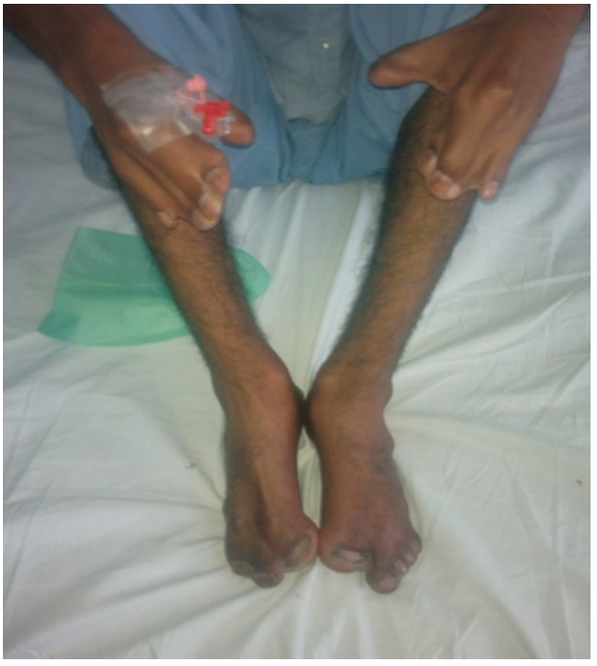

Figure 2. Symmetric polysyndactyly

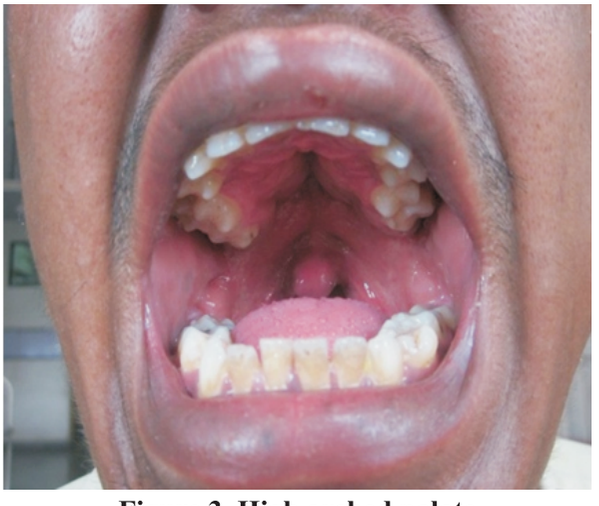

Figure 3. High arched palate

After obtaining a written informed consent from the patient and his relatives, intubation was planned in a spontaneously breathing anaesthetized patient. Anticipating a difficult airway all available airway management devices including laryngeal mask airway, flexible fibreoptic bronchoscope and flexible stylet were kept ready.The cricothyroid membrane was identified and marked prior to any airway manipulation. After intubation, we planned to keep the patient immobile by using nondepolarising muscle relaxants.

After securing a 20 gauge intravenous cannula on dorsum of right hand, pulse oximeter, non-invasive blood pressure and ECG monitors were placed and the patient was pre-medicated with injection midazolam 1 mg, injection pentazocine 18mg and injection glycopyrollate 0.2mg. After adequate pre oxygenation, patient was induced with injection thiopentonesodium 250mg. Manual in line traction andstabilization of the neck was maintained. Methylcellulose eye drops were instilled and left eye covered. As anticipated, mask ventilation was difficult because of the difficulty in lifting the jaw and inadequate mask fit due to the mid face hypoplasia, receding mandible, large tongue and a high arched palate along with a deviated nasal septum. An oropharyngeal airway was placed to assist ventilation to avoid airway obstruction. Sevoflurane was added to the gas mixture to deepen the plane of anaesthesia and improve mask ventilation.

At this point of time, we injected 40mg of injection lidocaine intravenously and did a laryngoscopy to evaluate the feasibility of intubation in a spontaneously breathing anaesthetized patient. As anticipated, the mouth opening was limited with a Cormack - Lehane laryngoscopic view of grade 3. The laryngeal apparatus was sprayed with 3 puffs of lidocaine and after optimal external laryngeal manipulation, the patient was intubated with a 7.5mm cuffed portex tube with a stillete. After confirming the tube placement, the patient was paralysed with injection vecuronium 2 mg and maintained on a mixture of isoflurane, oxygen and nitrous oxide. Rest of the surgical and anaesthetic course was uneventful. The effect of muscle relaxant was reversed and patient was extubated. Following extubation, patient had 2-3 episodes of vomiting which were managed by thorough oral suctioning and lowering the head end of the OT table. After observing the patient in OT for 30 minutes, the alert and conscious patient was shifted to the recovery room

Discussion

In Apert's syndrome, there is early synostosis of the spheno-occipital synchondrosis. This precipitates an especially short posterior cranial base resulting in the reduction of the pharyngeal height. The nasal height and depth are also decreased. The nasopharyngeal and oropharyngeal attenuation coupled with a potentially inflexible neck compound the problems in an already problematic airway. It becomes readily apparent that individuals with Apert's become mouth breathers out of necessity, due to reduced airway patency.

Apert's and Crouzon's syndrome seem to be the same syndrome, with the exception of syndactyly of hands and feet in Apert's syndrome. Both syndromes share a number of clinical and radiographic findings, such as premature synostosis of calvaria, maxillary hypoplasia, and mandibular overjet. However, cleft or pseudocleft palate is a frequent finding in Apert's syndrome, whereas these traits are extremely rare in Crouzon's syndrome [2].

The most prevalent concern is airway obstruction defects and management issues followed by altered respiratory mechanics. Cartilaginous abnormalities of the trachea, fusion of the cervical vertebrae, tracheal stenosis and angular deviation of the trachea may contribute to respiratory morbidity or difficult intubation in Apert's syndrome [3].

Proper positioning and supportive care of the patient's head during intubation and surgical or invasive procedures should prevent most spinal nerve injuries and possible paralysis in these patients. In addition, possible adverse reactions to certain medications that may cause cardiac arrhythmias, rigidity, hypertension, prolonged sedation, acute renal failure, and respiratory distress must be monitored in patients with specific genetic diseases. Approximately 60% of the reported 10,000 genetic conditions have central or peripheral nervous system abnormalities. A significant proportion of patients with Apert's syndrome have mental retardation. It is reported that 52% of the patients' IQ has been lower than 70. In these patients, significant social problems, speech difficulties, and attention deficit are noted [4]. In addition, hearing impairment and blindness are recognized features of several genetic diseases that raise additional problems for the health care provider [5].

Obtaining a good mask airway in these patients may be difficult owing to a poor seal from facial asymmetry, small chin, and decreased facial height. Several different mask types and sizes should be on hand to secure the best possible mask fit. Ideally, neuromuscular blockade monitoring should be done in these patients because of their variable body composition. Also, the washout of volatile anaesthetics mayoccur at a rapid rate owing to thelack of significant fat uptake during maintenance anesthesia, thereby minimizing subsequent release back into the circulation during emergence [6].

Tosun and Sener's study showed that Apert's syndrome was in parallel with G6PD deficiency. The hemolysis can be precipitated by oxidant drugs, fava beans, or intercurrent infection. Drugs that may induce hemolysis include sulphonamides, chloramphenicol, aspirin, acetaminophen, penicillin, and streptomycin. Therefore, the drugs that may potentially induce hemolysis as result of G6PD deficiency should be avoided in Apert's syndrome [7].

Approximately 10% patients with Apert's syndrome have cardiac conduction defects associated with early childhood mortality. Genitourinary anomalies namely hydronephrosis and cryptorchidism may be present in 9% of these patients.Central nervous system malformations and raised intracranial pressures along with increased risk of regurgitation of gastric contents have also been noted [8].

Another challenge for the anaesthetist is intravenous access. This can be made more difficult when one or more limbs are being operated on. As these children may return to theatre for repeat procedures, this issue can become increasingly more problematic. Eyes are particularly susceptible to damage due to inadequate lid closure. It is important to lubricate the eyes, and ensure they are also taped and padded. There are multiple limb abnormalities and care must be taken to avoid pressure points. Postoperatively, it is important to monitor for signs of airway obstruction.

Conclusion

It is important for anaesthesiologists, to recognize risk factors and potential complications before sedation or anesthesia in rare genetic syndromes like Apert's as repeated surgical corrective procedures are common in these patients and thus avoid the potential morbidity and mortality.

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