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Research Article

EMERGENCY SURGERY IN A CHILD WITH APERT SYNDROME: THE ANESTHETIC CHALLENGES

Himanshu Kumar., Deb Sanjay Nag*., Devi Prasad Samaddar and Seelora Sahu

Department of Anaesthesia and Critical Care, Tata Main Hospital, Jamshedpur 831001, India

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A B S T R A C T

Apert syndrome is an extremely rare congenital anomaly with acrocephalosyndactylia accompanying various other anatomical anomalies. This case report describes a 15 year old child with perforated appendix who presented for an emergency surgery. The Central Nervous System (CNS) and cervical spine anomalies in a child with Apert syndrome can give rise to unique anesthetic challenges. In an emergency scenario, other associated congenital anomalies need to be identified clinically and the anaesthetic plan needs to be modified accordingly.

Difficult intubation and bag mask ventilation needs to be anticipated and planned accordingly. Potential of tracheomalecia and bronchiectatic changes can give rise to ventilatory challenges in the intraoperative period. Opioids and other perioperative anaesthetic drugs can have a residual depressive impact on the CNS and lead to respiratory compromise. Awareness about the associated anomalies and preparedness for the known anesthetic challenges could minimize the risk to such patients.

INTRODUCTION

Apert syndrome is an extremely rare congenital anomaly that has been named after the French physician who first described the acrocephalosyndactylia syndrome in 1906. With a birth prevalence of 15.5/1,000,000 births, this autosomal dominant condition is caused by a two adjacent mutations (Ser252Trp & Pro253Arg) in the extracellular immunoglobulin (Ig) domains of fibroblast growth factor receptor 2 (FGFR2). [1,2] It is characterized by "coronal suture fusion, abnormal cranial base development, syndactyly of the hands and feet, symphalangism (fusion of digital phalanges), radio-humeral fusion, and varying degrees of neurocognitive impairment". [3] It is also associated with major cervical spine abnormalities in greater than 50% of the cases which include fusion of posterior elements, spina bifida occulta or atlanto-axial subluxation. [3]

Although Barnett S *et al* retrospectively reviewed the perioperative case notes of 61 children with Apert syndrome over a 14-year period, there are very few case reports discussing the anesthetic management in children with Apert syndrome. [4] A literature search was performed in PubMed in November 2016 for all published literature on the anesthetic management of Apert syndrome from 1979 to 2015. The search strings used for this research were: Apert syndrome [AND] case report [OR] case reports, [AND] anesthesia [OR] anaesthesia.

Corresponding author:* **Deb Sanjay Nag
Department of Anaesthesia and Critical Care, Tata Main Hospital, Jamshedpur 831001, India

The search yielded only 16 publications, but 3 among these were excluded for being other variants of acrocephalosyndactylia (2 cases of Saethre-Chotzen syndrome and one case of Carpenter syndrome) [13-15]. The 13 remaining case reports illustrates 15 cases (two publications reported two cases each) discussing the anesthetic management in children with Apert syndrome. While all reported cases were for elective surgeries where there is adequate time to review and investigate the child, we report, possibly the first case where a child with Apert syndrome presented for an emergency surgery.

Case Report

A 15 year child presented to the emergency department late in the evening with history of severe abdominal pain for the last 8 hours. He denied any other symptom apart from nausea. The abdominal pain was abrupt in onset, started with the right lower quadrant of the abdomen and subsequently become generalized. On examination, the child was febrile (100.6 degree Fahrenheit), had sinus tachycardia (on the ECG monitor, with a heart rate of 112/minute) and a Non Invasive Blood Pressure (NIBP) of 112/76 mm Hg. Abdominal examination revealed generalized tenderness all over the abdomen with guarding and rigidity in the right lower quadrant and periumbilical area. A surgical opinion was sought and abdominal ultrasound and an erect abdominal X–Ray were advised. While the X-Ray abdomen was unremarkable, the ultrasound finding suggested a perforated appendix with intraperitoneal extraluminal air locules, free

peritoneal fluid and surrounding bowel loops. The patient was planned for an emergency laparotomy and was evaluated by anesthesiologist.

The parents reported that the child had abnormal head size and fused digits of all four limbs since birth. They also mentioned that he was clinically diagnosed as Apert syndrome following delivery. Although the perinatal period was otherwise uneventful, the child used to have recurrent upper respiratory tract infections responsive to antibiotic therapy. However, he suffered from one episode of pneumonia needing hospitalization and intravenous antibiotics at the age of 6 years. Delayed milestones of development along with hearing impairment, snoring during sleep and daytime somnolence was also reported. There was no mention of seizures, cyanotic spells or any symptom related to cardiovascular disease. On examination, the child weighed 46kg, had brachycephaly with frontal prominence and a flattened occiput, low set ears, bilateral proptosis and depressed nasal bridge. There was fusion of the digits in all four limbs. The child's airway examination revealed adequate mouth opening, mottled discolored teeth with malocclusion, high arched palate and a modified Mallampati Grade 2 view. Neck movement was only mildly restricted. No added or abnormal sound was heard during auscultation of cardio-respiratory system. Fundoscopic examination of eye was also normal.

The child was premedicated with intramuscular glycopyrrolate 0.2 mg 45 minutes prior to shifting the child to the operating room. An intravenous access was established in the median cubital vein at the emergency department itself. Routine monitors were attached and baseline vitals were recorded in the operating room prior to induction. Anesthesia was induced with intravenous fentanyl 90 mcg, 100 mg propofol and after establishing the ability to ventilate with manual intermittent positive pressure ventilation with face mask, vecuronium 4 mg was given. Proper seal with a clear face mask was established with difficulty due to the abnormal facial anatomy, but adequate bag-mask ventilation could be confirmed by noting the chest-rise and end tidal carbon dioxide (ETCO2). Glottic visualization was adequate (Cormack–Lehane grade II) with a standard size 3 Macintosh blade and trachea was intubated with a 6.5 mm (ID) cuffed endotracheal tube. General anesthesia was maintained with intermittent doses of fentanyl and vecuronium along with isoflurane, nitrous oxide and oxygen by the circle system. The surgery for the perforated appendix lasted for 85 minutes and neuromuscular blockade was reversed with glycopyrrolate and neostigmine. Tracheal suctioning was done in a deeper plane of anesthesia and tracheal extubation was done only when the child was fully awake and could respond to verbal commands. Postoperative analgesia was maintained with intramuscular diclofencac sodium and intermittent doses of intravenous tramadol. The child recovered over the next few days and was discharged on the 7th post-operative day.

DISCUSSION

The presence of Central Nervous System (CNS) and cervical spine anomalies in a child with Apert syndrome can give rise to unique anesthetic challenges. It is also often associated with other congenital cardiac (overriding aorta, septal defects), genitourinary (polycystic or hydronephrotic kidneys) or pulmonary (tracheal stenosis or pulmonary hypoplasia) anomalies which necessitates additional workup and cross-consultations to rule out their presence. [5] While it is possible in an elective scenario, in an emergency situation, the anesthesia plan needs to be based on the clinical findings and readily available laboratory investigations.

Apert syndrome is often associated with enlarged ventricles, "crowded foramen magnum", and absence of septum pellucid or agenesis of corpus callosum. [3] Our patient presented only with delayed milestones. Complains of snoring and daytime somnolence led us to suspect obstructive sleep apnea (OSA) in the child. Pijpers M noted that in children with syndromal craniofacial synostosis, the actual incidence of obstructive sleep apnea was as high as 53% as compared to a suspected incidence of 26%. [6] Difficult intubation and bag mask ventilation is anticipated in such patients due to the presence of various upper airway and facial abnormalities (midface hypoplasia, small naospharynx, choanal atresia, high arched palate). Difficult intubation due to trismus has also been reported in Apert syndrome. [7] As these children are prone to rapid airway obstruction and oxyhaemoglobin desaturation during induction, the anaesthesiologist should be ready with alternative airway management plan and an armamentarium of airway adjuncts. [8] Though, in our case acceptable seal with a clear face mask could only be achieved with difficulty, tracheal intubation was relatively simple due to adequate glottic visualization (Cormack–Lehane grade II).

Major cervical spine abnormalities including fusion of posterior elements and multilevel fusion are present in over 50% of the patients with Apert Syndrome. [3] However, as majority of these anomalies are at the level of lower cervical spine, they are of lesser significance for intubation as head and neck positioning for intubation primarily involves the movement at the level of upper cervical spines. [9] Apert syndrome has been associated with tracheal anomalies like "partial cartilage sleeve" causing respiratory compromise and retained secretions. [10] Tracheomalacia with a false carina leading to a blind pouch causing compromised ventilation after intubation has also been reported. [11] These patients often have bronchiectatic changes leading to recurrent respiratory infections. Although our patient gave a history of recurrent respiratory infections, her chest auscultation was did not reveal any abnormal finding. We prophylactically used glycopyrrolate to decrease secretions. No respiratory compromise was encountered in our patient during the intraoperative or immediate postoperative period.

Intravenous access is expected to be difficult in patients with syndactyly. However, in our patient it was established in the median cubital vein without any problem.

The central nervous system manifestations of Apert Syndrome as well as OSA make these patients prone to abnormal effects of residual anesthetics agents and opioids in the immediate post-operative period. The patients should be continuously monitored for apnea or hypopnea and episodes of oxyhaemoglobin desaturation or hypercapnia. They may need non-invasive positive pressure ventilation in the form of Continuous Positive Airway Pressure (CPAP) or Bilevel Positive Airway Pressure (BiPAP) post-operatively. However, in our patient, tracheal extubation was done when the patient was fully awake and he was managed with supplementary oxygen by face mask. Non-invasive ventilatory support was not needed to maintain adequate respiration. The use of non-

steroidal anti-inflammatory drugs could reduce the use of opioids and any respiratory depression. The use of tramadol which rarely causes "clinically relevant respiratory depression" in equianalgesic doses as compared to morphine further enhances perioperative safety in such patients. [12] Therefore, we used tramadol as the post-operative analgesic and further supplemented this with diclofenac.

CONCLUSION

Being a rare congenital anomaly associated with significant craniofacial and visceral anomalies, the anesthetic management of such patients needs a multidisciplinary evaluation and radiological screening craniofacial, cervical spine and chest), ultrasonography (visceral) and echocardiography prior to an elective surgery. However, in an emergency scenario, awareness about the associated anomalies, clinical evaluation to specifically look for them and preparedness for the known anesthetic challenges could minimize the risk to such patients.

References

1. Cohen MM Jr, Kreiborg S, Lammer EJ, Cordero JF, Mastroiacovo P, Erickson JD, Roeper P, Martínez-Frías ML.Birth prevalence study of the Apert syndrome. *Am J Med Genet*. 1992 Mar 1; 42(5):655-9.
2. Wilkie AO1, Slaney SF, Oldridge M, Poole MD, Ashworth GJ, Hockley AD, Hayward RD, David DJ, Pulleyn LJ, Rutland P, *et al*. Apert syndrome results from localized mutations of FGFR2 and is allelic with Crouzon syndrome. *Nat Genet*. 1995 Feb; 9(2):165-72.
3. Breik O, Mahindu A, Moore MH, Molloy CJ, Santoreneos S, David DJ. Central nervous system and cervical spine abnormalities in Apert syndrome. *Childs Nerv Syst.* 2016 May; 32(5):833-8. doi: 10.1007/ s00381-016-3036-z. Epub 2016 Feb 10.
4. 4. Barnett S1, Moloney C, Bingham R.Perioperative complications in children with Apert syndrome: a review of 509 anesthetics. *Paediatr Anaesth*. 2011 Jan; 21(1):72-7. doi: 10.1111/j.1460-9592.2010.03457.x. Epub 2010 Nov 15.
5. Cohen MM Jr1, Kreiborg S.Visceral anomalies in the Apert syndrome. *Am J Med Genet*. 1993 Mar 15; 45(6):758-60.
6. Pijpers M1, Poels PJ, Vaandrager JM, de Hoog M, van den Berg S, Hoeve HJ, Joosten KF. Undiagnosed obstructive sleep apnea syndrome in children with syndromal craniofacial synostosis. *J Craniofac Surg*. 2004 Jul; 15(4):670-4.
7. Morris GP, Cooper MG.Difficult tracheal intubation following midface distraction surgery. *Paediatr Anaesth.* 2000; 10(1):99-102.
8. Patino M, Sadhasivam S, Mahmoud M.Obstructive sleep apnoea in children: perioperative considerations. *Br J Anaesth*. 2013 Dec; 111 Suppl 1:i83-95. doi: 10.1093/bja/aet371.
9. Kumar N, Arora S, Bindra A, Goyal K.Anesthetic management of craniosynostosis repair in patient with Apert syndrome. *Saudi J Anaesth* 2014:8:399-401
10. Cohen MM Jr, Kreiborg S.Upper and lower airway compromise in the Apert syndrome. *Am J Med Genet*. 1992 Sep 1; 44(1):90-3.
11. Hutson LR Jr, Young E, Guarisco L. Tracheal anomalies complicating ventilation of an infant with Apert syndrome. *J ClinAnesth*. 2007 Nov; 19(7):551-4.
12. Houmes M, Voets MA, Verkaaik A, Erdmann W, Lachmann B. Efficacy and safety of tramadol versus morphine for moderate and severe postoperative pain with special regard to respiratory depression. *Anesth Analg* 1992; 74:510-4.
13. Easely D, Mayhew JF.Anesthesia in a child with Saethre-Chotzen syndrome. *Paediatr Anaesth*. 2008 Jan; 18(1):81.
14. Sharma A, Patel N, Arora S, Ramachandran R. Child with Saethre-Chotzen syndrome: anesthetic management and literature review. *Acta Anaesthesiol Belg*. 2014; 65(4):179-82.
15. Bhardwaj M, Grange C.Caesarean section in a parturient with Carpenter syndrome and corrected Tetralogy of Fallot. *Int J Obstet Anesth*. 2013 Jul; 22(3):251-4. doi: 10.1016/j.ijoa.2013.04.009.

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