International Journal of Medical and Health Research
ISSN: 2454-9142
www.medicalsciencejournal.com
Volume 4; Issue 4; April 2018; Page No. 91-93

Anesthesia and airway management in a case of Apert syndrome

Deeksha Bindal, Varun Kumar Saini, Dr. Satyendra Patidar*
Department of Anesthesiology, Critical Care and Pain Management, Govt. RDBP Jaipuria Hospital, Jaipur, Rajasthan, India

Abstract
Apert syndrome is a rare autosomal dominant developmental malformation characterized by craniofacial and limb deformities synostosis, symmetric severe syndactyly and a variety of abnormalities of the skin, skeleton, brain, and visceral organs. We share our experience of anesthetic management of an infant with apert syndrome posted for syndactyly release.

Keywords: Apert syndrome, difficult mask ventilation, difficult intubation, difficult intravenous cannulation, syndactyly

Introduction
Apert syndrome or acrocephalosyndactyly is a rare autosomal dominant developmental malformation characterized by craniosynostosis, a cone shaped calvarium, midface hypoplasia, pharyngeal attenuation, ocular manifestations and syndactyly of the hands and feet [1], abnormality of skin, skeleton, brain and visceral organs.
Mutations of either Ser252Trp or Pro253Arg in fibroblast growth factor receptor 2 (FGFR2) are responsible for nearly all known cases of Apert syndrome [2]. The inheritance is autosomal dominant, with a frequency of 1 in 100,000-160,000.
During anesthesia, difficult intubation and ventilation may be observed because of abnormal airways. Children with craniofacial anomalies often have abnormal airways. Apert syndrome is associated with the loss of lower and upper airway patency as well as respiratory complications during general anesthesia [3, 4] Intubation and ventilation may be difficult during general anesthesia [3, 5, 6]

Case report
A 3 month old male baby presented to orthopaedics OPD with complaints of atypical facial appearance and bilateral symmetrical syndactyly of all digits of hand and feet since birth and was scheduled for reconstruction surgery of rt. hand and rt foot. The infant was born at 36 wks of gestational age out of an uneventful pregnancy and through normal vaginal delivery. At the time of his birth, his mother was 20 yrs and his father was 30 yrs. his father also has same symptoms since birth. (His father was born out of second degree consanguinous marriage,when his grandfather was 55 yrs of age).

On examination
The boy's skull appeared brachycephalic with a flat occiput.
1. Flat facies, shallow orbits, proptosis, slight hypertelorism present
2. Maxillary hypoplasia, micrognathia, large tongue.
3. Bilateral symmetrical cutaneous syndactyly of all digits of both hands and feet.
4. He had difficulty with feeding, malnutrition and respiratory problems since birth, for which he had been treated several times in the pediatric intensive care unit.

Airway Examination
Incomplete nasal obstruction.
Maximum mouth opening as shown in figure
Grade 3 intubation score.
The thyrohyoid distance and the atlanto-occipital joint movement were normal.
The following investigations were done :
1. An X-ray of the head revealed craniosynostosis of the coronary suture.
2. No structural and functional abnormality of heart was observed in echocardiogram.
3. Cranial, abdominal, and urinary system ultrasonographies were normal.
4. Molecular genetic analyses could not be done.

Pre-operative laboratory findings were normal. After 4 h of fasting, premedication consisted of intramuscular (im) midazolam (0.1 mg/kg) and atropine (0.015 mg/kg). ECG and SpO2 were monitored. Since we anticipated difficult intubation, the anesthetic induction occurred with sevoflurane in nitrous oxide/oxygen (65%-35%) via a facemask followed by laryngoscopic oral intubation without neuromuscular blockers. A 3-mm internal diameter (ID) endotracheal tube with an inserted curved guide wire was passed at the third laryngoscopy. Anesthesia was maintained with 2%sevoflurane in nitrous oxide/oxygen. During anesthesia, there was wheezing, and clear secretions were frequently aspirated from the endotracheal tube. The patient was extubated after surgery with no anesthetic complications.

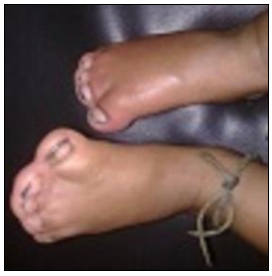

Fig 1

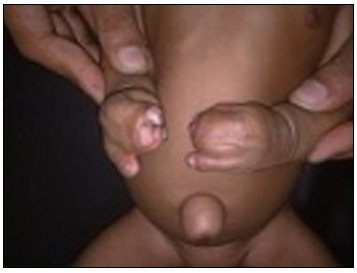

Fig 2

Fig 3

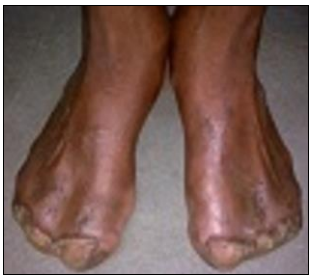

Fig 4: Apert syndrome

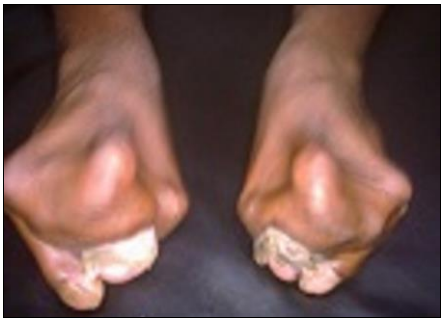

Fig 5

Discussion

Apert syndrome or acrocephalosyndactyly is a rare malformation syndrome first described by Wheaton in 1894 and later by Apert in 1906 [7].

It occurs with a reported birth prevalence of 1/65 000 [8].

The hallmarks of the syndrome include craniosynostosis (abnormal development and premature fusion of the cranial sutures), symmetric severe syndactyly (cutaneous and bony fusion of the digits), and a variety of abnormalities of the skin, skeleton, brain, and visceral organs. The condition results from a specific missense mutation in the gene-encoding fibroblast growth factor receptor-2 (FGFR-2), mapped to 10q26 chromosome. This pleiotropic gene is involved in the complex intercellular signalling network that controls cell proliferation, differentiation, migration, and survival in many different contexts, including embryonic development, angiogenesis, and malignancy [9].

Mutations of the FGFR-2 gene have also been associated with several other craniosynostosis malformation syndromes, including Crouzon,

Jackson–Weiss, Pfeiffer, and Beare–Stevenson cutis gyrate syndromes [10].

Most cases of Apert syndrome are sporadic, while autosomal dominant transmission and germinal mosaicism have also been reported [11].

The paternal origin of new mutations, specifically related to age effect, has been elucidated [12].

Premature fusion of cranial sutures, most commonly of the coronal suture, is observed in all patients with Apert syndrome. Antero–posterior shortening of the cranial base leads to acrocephaly or brachycephaly. Other characteristic craniofacial abnormalities include prominent forehead with skin wrinkling, broad cranium, and a flat occiput. Hypertelorism, proptosis, and strabismus are often present due to shortening of the bony orbit [13].

Additional craniofacial features include a short, broad nose with a bulbous tip, micrognathia, and a cleft palate [14].

Symmetric syndactyly of the hands and feet is another universal finding in patients with Apert syndrome. Centralnervous system abnormalities include defects of the corpuscallosum and limbic structures, ventriculomegaly, and progressive hydrocephalus. A significant number of patients function at an intellectual level two standard deviations below the mean [15].

Cardiovascular and genitourinary defects occur in 10% and 9.6% of patients with the syndrome, respectively [16]

A number of cutaneous manifestations are characteristic of Apert syndrome [17].

Anesthetists should be aware that airway dysmorphism carries a risk of difficult intubation and respiration problems in patients with Apert syndrome [18]. Craniofacial anomalies are often associated with airway obstruction, especially during sleep, and can cause obstructive sleep apnea [19, 20]. However, preoperative evaluation showed airway or other problems in our patient. Elwood *et al*. [21]. Found respiratory complications in 33% of patients with Apert syndrome. As in our case, upper and lower respiratory infections were due to respiratory complications from the underlying condition, particularly increased secretions [18, 20].

The following classical features should be evaluated preoperatively:

- High Mallampati score
- Reduced thyromental distance
- Reduced sternomental distance
- Restricted atlanto-occipital joint movement
- Restricted temporo-mandibular joint function
- Limited mouth opening

Intubation aids, such as Combitube, laryngeal masks and emergency tracheotomy sets must be available.

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 syndrome [20] Tracheotomy is difficult in children and may lead to complications; percutaneous cricothyroidotomy is less likely to be required in children with craniofacial anomalies, such as Apert syndrome [21, 22]. Fiberoptic and retrograde intubations, which are difficult in children, have been used in cranio-facial anomalies [19, 21]. Visceral anomalies may also cause complications.

We prepared laryngeal masks and emergency tracheostomy sets for our case, although neither was needed. However, in Apert syndrome, the anesthetist must be ready for airway problems, intubation difficulties and even visceral anomalies.

In particular, Apert syndrome with visceral anomalies may cause significant problems for anesthesia and surgery. In addition, if difficult airway management is a concern, the induction of the anesthesia should be administered with inhalation agents and without neuromuscular blockers to maintain spontaneous ventilation. For these reasons, thorough pre-operative evaluation and planning is essential.

References

1. Batra P, Duggal R, Parkash H. Dentofacial characteristics in Apert, syndrome: a case report. J Indian Soc Pedod Prev Dent. 2002; 20:118-23.
2. Chen L, Li D, Li C, *et al*. A ser250trp substitution in mouse fibroblast growth factor receptor 2 (FGFR2) results in, craniosynostosis. Bone. 2003; 33:169-78.
3. Elwood T, Sarathy PV, Geiduschek JM, Ulma GA, Karl HW. Respiratory complications during anaesthesia in Apert syndrome. Pediatr, Anaesth. 2001; 11:701-3.
4. Pardos PC, Pazos JC. Airway management in an infant with Apert's syndrome. Rev Esp Anestesiol Reanim. 2000; 47:97-8.
5. Morris GP, Cooper MG. Difficult tracheal intubation following midface distraction surgery. Pediatr Anaesth, 2000; 10:99-102.
6. Lee YH, Liou SC, Chen C, Wong TK, Tan PP. Airway obstruction in general anesthesia two different episodes in the same patient: case report. Changgeng Yi Xue Za Zhi. 1999; 22:682-6.
7. Apert M. De l'acrocephalosyndactylie. Bull Mem Soc Med Hop Paris. 1906; 23:1310-1313.
8. Cohen MM Jr, Kreiborg S, Lammer EJ, *et al*. Birth prevalence study of the Apert syndrome. Am J Med Genet. 1992; 42:655-659.
9. Wilkie AO, Slaney SF, Oldridge M, *et al*. Apert syndrome results from localized mutations of FGFR2 and is allelic with Crouzon syndrome. Nat Genet. 1995; 9: 165-172.
10. Hollway GE, Suthers GK, Haan EA, *et al*. Mutation detection in FGFR2 craniosynostosis syndromes Hum Genet. 1997; 99:251-255.
11. Allanson JE. Germinal mosaicism in Apert syndrome. Clin Genet.1986; 29:429-433.
12. Moloney DM, Slaney SF, Oldridge M, *et al*. Exclusive paternal origin of new mutations in Apert syndrome. Nat Genet. 1996; 13:48-53.
13. Steffen C. Acneiform eruption in Apert syndrome. Acrocephalosyndactyly. Arch Dermatol. 1982; 118:206-208.
14. McNaughton PZ, Rodman OG. Apert syndrome. Cutis. 1980; 25:538-540.
15. Cohen MM, Jr Kreiborg S. The central nervous system in the Apert syndrome. Am J Med Genet. 1990; 35:36-45.
16. Cohen MM, Jr Kreiborg S. Visceral anomalies in the Apert syndrome. Am J Med Genet1993; 45:758-760.
17. Cohen MM, Jr Kreiborg S. Cutaneous manifestations of Apert syndrome. Am J Med Genet. 1995; 58:94-96.
18. Pardos PC, Pazos JC. Airway management in an infant with Apert's syndrome. Rev Esp Anestesiol Reanim. 2000; 47:97-8.
19. Kaymak C, Gulhan Y, Ozcan AO, Safak MA, Oguz H. Anaesthetic approach in a case of Goldenhar's syndrome. Eur J Anaesthesiol. 2002; 19:836-8.
20. Perkins JA, Sie KCY, Milczuk H, Richardson MA. Airway management in children with craniofacial anomalies. Cleft Palate Craniofac J. 1997; 34:135-140.
21. Elwood T, Sarathy PV, Geiduschek JM, Ulma GA, Karl HW. Respiratory complications during anaesthesia in Apert syndrome. Pediatr Anaesth. 2001; 11:701-3.
22. Morris GP, Cooper MG. Difficult tracheal intubation following midface distraction surgery. Pediatr Anaesth. 2000; 10: 99-102.