Case Report

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Anaesthetic Management of a Patient with Apert Syndrome

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Abstract

Introduction: Apert syndrome is a type of acrocephalosyndactilia that includes several abnormalities. Number of coexisting conditions should be taken under consideration while giving anaesthesia. We report our experience in the anaesthetic management of case of Apert syndrome, referred to us for syndactyly release in both upper limbs. *Case Report:* A 13-months male child with Apert syndrome, weighing 7.29 kg, scheduled for syndactyly release of both upper limbs (fusion of digit 2 to 4), had facial dysmorphism with macroglossia, craniosynostosis, short nose with widely depressed nasal bridge with bulbous tip, proptosis, wide set eyes, mandibular retraction, shield chest deformity (pectus excavatum), limited mouth opening (M.P. grading III), high arched palate, history of sleep apnoea, patent foramen ovale with left to right shunt, brachycephaly and mild ventriculomegaly. *Anaesthetic Management:* Case was managed under general anaesthesia with intubation in spontaneously breathing anaesthetised patient without neuromuscular blocking agents. Awake extubation was done with no intra-operative or post-operative complications. *Discussion:* Anaesthetic management of a patient with Apert syndrome may be challenging. The anaesthesiologist must be ready for airway problems, intubation difficulties, obtaining a good mask seal, difficult IV access. Raised intra cranial pressure, sleep apnea, possibility of mental retardation and other associated anomalies should always be considered. *Conclusion:* Ever vigilant Anesthesiologist who has armoured himself with all the potential problems likely to arise, in the management of children with this rare genetic syndrome (who may need repetitive surgeries), will make all the difference in the successful outcome of these patients.

Keywords: Apert Syndrome; Acrocephalosyndactilia; Macroglossia; Craniosynostosis.

Introduction

Apert syndrome, named after the French paediatrician Eugene Apert, is a congenital autosomal dominant disease with an incidence of 1 per 160,000 live births that affects both males and females equally [1]. Mutation of 'Fibroblast Growth Factor Receptor 2' (FGFR 2) on chromosome number 10 gene has been observed [2]. The syndrome is characterized by bracycephaly, craniosynostosis, midface hypoplasia, hypertelorism, choanal stenosis, multidigit hand and foot syndactyly. Associated anomalies are rare and may include cardiac defects, polycystic kidneys and pyloric stenosis [1]. The cervical spine is fused in 68% of those with Apert syndrome, usually at C5-6. Upper airway obstruction is present due to nasopharyngeal deformity and choanal atresia. Almost 50% also suffer from obstructive sleep apnea which may lead to corpulmonale [3].

Case Report

A 13-months male child with Apert sydrome, weighing 7.29 kg was scheduled for syndactyly separation of both upper limbs (fusion of digit 2 to 4).

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A written consent from the patient's mother to publish the case was obtained. Following were noted during Pre-Anaesthetic examination- facial dysmorphism with macroglossia, craniosynostosis evidenced by peaked vertical elongated head, short nose with widely depressed nasal bridge with bulbous tip, proptosis, wide set eyes and mandibular retraction, symmetric syndactyly of hands and feet, shield chest deformity (pectus excavatum) with widely and inferiorly placed nipples, with limited mouth opening and high arched palate. Parents gave a history of sleep apnoea, with no history of respiratory tract infections. On general examination, his pulse rate was 138/min with regular rhythm, respiratory rate was 26/min, Mallampatti grading was III. Systemic examination was essentially normal. Routine investigations including X-ray chest, skull, dorso-lumbar spine and USG Abdomen did not show any gross anomaly. 2D Echo showed patent foramen ovale with left to right shunt, whereas MRI Brain revealed brachycepahly and mild ventriculomegaly.

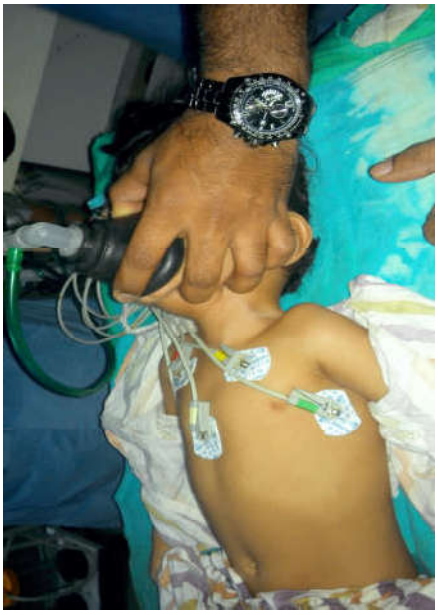


Fig. 4

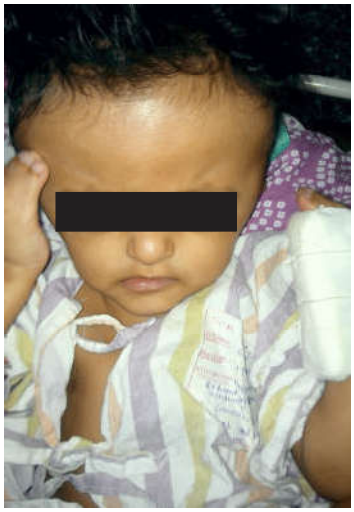
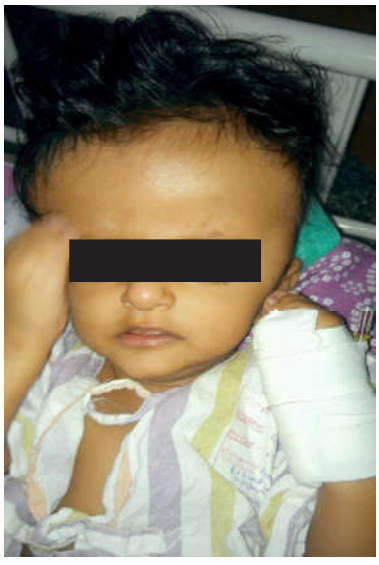


Fig. 1:

Fig. 4



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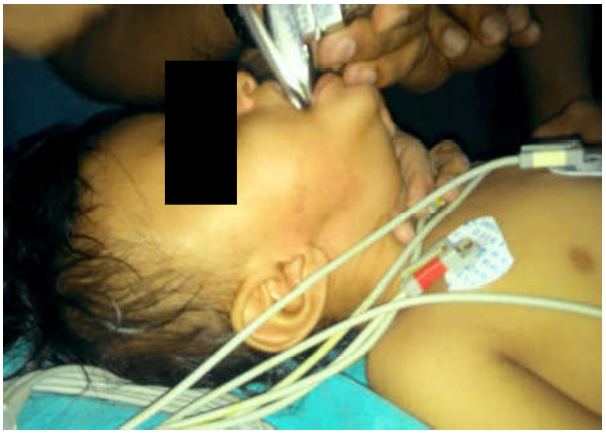

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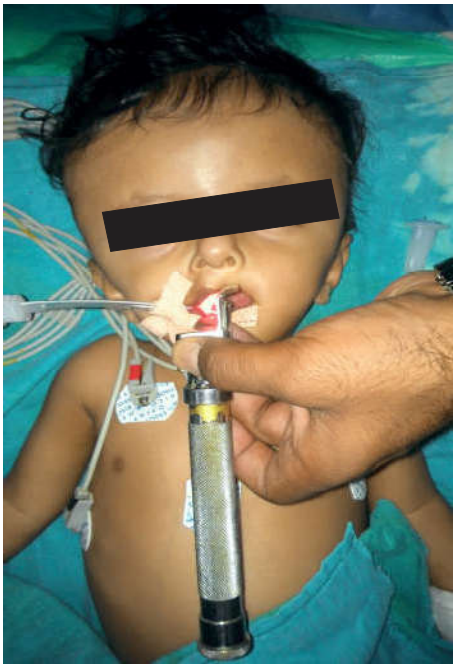

Fig. 6

Anaesthetic Management

Patient was accepted under ASA-III and a written informed consent to publish the case was obtained from parents. Anticipating a difficult airway, intubation was planned in spontaneously breathing anaesthetised patient with all emergency airway devices kept as standby.

After securing a 24-gauge intravenous cannula over Rt. Foot, pre-medication was done in presence of his parents, with injection Glycopyrrolate 0.04mg, injection Fentanyl 5µg, and injection Midazolam 0.02mg intravenously. In operation theatre, pulse oximeter, non-invasive blood pressure and ECG monitors were attached, preoxygenation done with 100% oxygen by mask for 3 minutes. Neck stabilization done with bilateral support. As anticipated, mask ventilation was difficult due to mid facial hypoplasia, receding mandible, large tongue and high arched palate. Jaw thrust and obtaining adequate mask fit was difficult. Guedel's airway was placed to assist ventilation and to avoid airway obstruction. Induction was done with Inj.Propofol 15mg itravenously. Sevoflurane and Nitrous-oxide was added to oxygen, to deepen the plain of anaesthesia and improve mask ventilation.

Endotracheal intubation was done on second attempt after optimal external laryngeal manipulation, using un-cuffed portex tube, number 2.5 with stillete. After confirming the tube placement and fixing it, patient was maintained on sevoflurane, nitrous oxide and oxygen by spontaneous-assisted ventilation through J-R Circuit. Intraoperatively injection Fentanyl (2+2+2+2) 8 µg and inj. Paracetamol 100mg diluted in 10cc normal saline were given intravenously for analgesia. Rest of the surgical and anaesthetic procedure was uneventful. Awake extubation done after surgery with no anaesthetic complications and observed in recovery room for 30 minutes after gaining complete consciousness. 170 mL crystalloid were given in this 2.5-hour case.

Dicussion

Anaesthetic management of children with Apert's syndrome is very challenging not only because of the characteristic features like brachycepahly, midface hypoplasia and orbital hypertelorism causing difficult airway management but also because of the associated congenital defects like cleft palate (30% cases), congenital heart disease (10%) and genito-urinary abnormalities (10%) [4]. Severe synostosis can result in raised intracranial pressure and developmental delay. The main concern of the anaesthesiologist is to ensure normoxia and normocarbia in the entire perioperative period. Syndactyly with fusion of 2 to 4 digits can make iv access very difficult especially when bilateral correction is planned in the same sitting.

A significant proportion of patients with Apert's syndrome have mental retardation. It is reported that 52% of patients IQ is lower than 70 leading to difficulty in administration of premedication and induction of anaesthesia [5]. Parental presence during these periods is definitely beneficial. Thorough pre-anaesthetic assessment to recognize the presence and severity of airway obstruction which would be worsened by loss of consciousness is essential.

Obtaining a good mask seal in these patients may be difficult owing to facial asymmetry, small chin, and decreased facial height. Several different mask types and sizes should be kept ready to secure the best possible mask fit. Limited mouth opening, restricted temporo-mandibular joint movement and atlanto-occipital joint movement poses potential hazards of intubation.

Intra-operatively eyes are susceptible to damage due to partial lid closure. So, eyes should be lubricated and also tapped and padded. There are multiple limb abnormalities and care must be taken to avoid pressure points.

Conclusion

If difficult airway management is a concern, the induction of anaesthesia should be considered with inhalational agents without neuromuscular blockers and whenever possible, regional anaesthesia should be preferred.

Ever vigilant anaesthesiologist who has armoured himself with all the potential problems likely to arise, in the management of children with this rare genetic syndrome (who may need repetitive surgeries), will make all the difference in the successful outcome of these patients.

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