Anaesthetic Considerations in an Infant with Cryptophthalmos – Anophthalmos

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Abstract: Several instances of Cryptophthalmos-Anophthalmos syndrome, including Fraser's syndrome have been documented and the various anomalies found associated have been known, including the anaesthetic complications in cardiac as well as other major surgeries. Here we discuss the anaesthetic management and considerations in a non-cardiac case, including the difficult airway, for which retro-molar technique of intubation was successfully performed after one unsuccessful direct laryngoscopy intubation attempt.

CASE REPORT

Cryptophthalmos meaning 'hidden eye' refers to a condition where the eye is malformed and maybe completely covered by skin. (1) This is found in about 88% population affected by Fraser syndrome also known as Fraser-Francois syndrome, Meyer-Schwickerath syndrome and Ulrich-Feichtiger syndrome among other names. (1)

The incidence of Fraser syndrome is 0.043 per 10,000 live born infants and 1.1 in 10,000 stillbirths. (2) Fraser syndrome is the result of various genetic mutations including mutations in FRAS1, FREM1, FREM2 and GRIP1 genes. Depending on the genes involved, other defects may be observed in the patient.

Systemic defects such as bilateral renal agenesis (45%), bilobed lungs, hypoplastic bladder, pulmonary hypoplasia and hydrocephalus have been commonly reported. Intersex development anomalies in genitals have also been reported such as micropenis or clitoromegaly, internal genitalia agenesis (streak ovaries), imperforate anus and cryptorchidism. (3)

Defects may include laryngeal deformities such as laryngeal and tracheal stenosis, choanal atresia, deformed nasal and oral anatomy and cranio-facial dysmorphism. (4)

As anaesthetists we should keep the above in mind while anaesthetising patients with this condition, and also be ready for any possible complication such as pneumoperitoneum due to cecal rupture caused by colonic atresia. Such colonic atresia also has propensity to lead to increased aspiration risk. (2)

A 23-day-old neonate, was referred to us in view of fused eyelids and deformity in eyes since birth.

Mother had history of non-consanguineous marriage, two miscarriages, both in first trimester. Uneventful natal history of present infant.

Diagnosed as ?Fraser syndrome - Left eye Anophthalmos, Right eye Cryptophthalmos and Right undescended testes. Associated with developmental delay. At two months of age, B scan both eyes confirmed ophthalmic findings.

Planned for Left upper eyelid formation with amniotic membrane graft / buccal mucosal graft under general anaesthesia, performed at 4 months of age.

On airway examination, mouth opening was adequate with MPC 1, dentation normal for age, and no mass/ distortion present orally. However, despite this, high risk consent was obtained due to known airway co-anomalies. 2DECHO screening was within normal limits.

Patient was shifted to OT, all monitor readings were within normal limits for age. One 22G cannula was secured on right forearm. Advanced airway devices including stillet and bougie were kept ready. Induction perfomed with Inj. Fentanyl 25 mcg and Inj. Atracurium 5mg. IPPV was done via bag and mask, which was adjusted to fit the face using two gauze pieces (due to depressed nasal bridge). Laryngoscopy was performed using Miller's blade size 2, however the glottic opening could not be clearly visualised. First intubation attempt was unsuccessful. Next, retro-molar technique was attempted from the right side and patient was successfully intubated with uncuffed PVC ETT size #4/10, fixed in place. All the IV cannulas were secured properly and all the bony prominences were padded adequately. The procedure lasted for 3 hours.

As backup airway, supraglottic airway device had been arranged for emergency use. Video laryngoscope was available but the appropriate size blade according to the patient's airway was not.

Care was taken to maintain airway patency and other physiologic parameters during the surgery. No fluctuations in any hemodynamic parameters were observed during the course of the operation and no untoward events such as laryngospasm took place during extubation.

Patient was successfully extubated and shifted to the recovery room. Postoperative period also remained uneventful till discharge.

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Airway security during syndromic cases is of prime importance due to known associations of laryngeal/tracheal anomalies along with the visible anomaly in Fraser's.

Retromolar technique of intubation is an alternative to regular intubation in difficult intubation cases. Here, the space behind the molar tooth of the lower jaw is utilised and the tube is passed behind it.

Since not all cases are associated with Cryptophthalmos or anophthalmos, presence of other isolated minor markers like Syndactyly, nasal coloboma or notching at tongue tip (1) should also raise suspicion and warrant a full workup.

Tracheal stenosis/ laryngeal stenosis (in about 30%) / choanal narrowing/ orofacial clefting/ craniofacial dysmorphism/ microstomia or micrognathia/ retrognathia and other airway abnormalities maybe present that have been documented to lead to difficult intubation and may even precipitate laryngospasm in the patient. (1)

M Saito et al decided to postpone the operation until their paediatric patient could cry fully without cyanosis when they encountered a similar case. Jagtap SR et al performed a retrograde intubation in their paediatric Fraser's syndrome case when they encountered difficult intubation.

Concurrent renal abnormalities such as bilateral or unilateral agenesis, cystic dysplasia of kidney, and hypoplastic bladder must be ruled out due to their documented association with Fraser syndrome.

Advanced airway devices such as stillet, bougie, and fiberoptic apparatus for retromolar intubation, must be kept ready, along with experienced anaesthesiologists on alert. Video-laryngoscopes also can be used to intubate the patient if all else fails.

Retromolar intubation is a non invasive technique of intubation wherein the space behind the last molar on either side is utilised to insert the endotracheal tube. Adequacy is determined beforehand and if required, a smaller size tube is inserted.

Regular monitoring of the vital parameters is a must to ensure that adequate ventilation and hemodynamic stability is being maintained. Cardiac anomalies must be ruled out pre-op by 2DECHO screening tests, as these are associated with high frequency, including left ventricular hypertrophy, coarctation of aorta, Ebstein anomaly variant, atrial septal defect and trunks arterioles with ventricular septal defect. (1)

Specific airway care must be extended till extubation and the post-operative period to ensure no laryngospasm, or lung collapse occurs, compromising ventilation.

In conclusion, Fraser syndrome usually presents with Cryptophthalmos, anoththalmos and Syndactyly, but may present at times without any physical anomalies, such as absent Syndactyly in our case. Due to this unique challenge we must be alert and actively search such cases for associated deformities and perform a thorough examination and investigations to systematically rule out any and all known co-anomalies especially those related to the airways and vital structures.

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