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CLINICAL REPORT

Fraser Syndrome: A Stumbling Block for the Anaesthesiologist

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Abstract Fraser syndrome is a rare autosomal recessive disorder with spectrum of malformations. Gamut of abnormal airway includes high arched palate, laryngeal atresia, hypoplasia, laryngeal stenosis etc. Laryngeal intubation difficulties have often been reported in literature. Airway management and decision process leading to emergency tracheostomy in these children has been discussed.

Keywords Paediatric · Fraser syndrome · Difficult intubation · Subglottic stenosis

Fraser syndrome is a rare autosomal-recessive inherited congenital disorder classically presenting with cryptophthalmos, ear and facial abnormalities, cutaneous syndactyly and genital malformations classical Fraser Syndrome is caused by mutation of the FRAS1 gene located on chromosome 4 at 4q21.21 [1]. Mutations of other genes FREM1, FREM2 and GRIP1 genes can also cause a similar clinical phenotype to Fraser syndrome [2]. The complete syndrome was described by Fraser [3].

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We report a case of Fraser syndrome with difficult airway for emergency tracheostomy.

A 3.5 kg 4 month old female child presented with respiratory distress in the emergency department with complaint of noisy breathing since birth which was not relieved with change in position. The pulse rate was 158/min, respiratory rate was 48/min with chest retractions, CRT was 2 s. Motor activity was normal and symmetrical. Her SpO_2 was 96% on room air. Child was pink, warm and well perfused.

Primary management in emergency in the form of oxygen by face mask, oro-pharyngeal suctioning, nebulisation with adrenaline 1:1000 and propped up position was initiated. Despite the above treatment and in view of worsening respiratory distress a decision for direct laryngoscopy and tracheal intubation was made. After two failed attempts, ENT opinion was sought and a cricoid split procedure followed by emergency tracheostomy in case of failure was planned.

The past history disclosed that the patient was born after an uncomplicated prenatal course at 35 weeks of gestation with 2.0 kg birth weight. The child was found to have cryptophthalmos left eye (hidden eye) and syndactyly in right upper limb at birth. There was no parental consanguinity and the older sibling was normal. The baby had delayed milestones as per age. A similar past episode of stridor revealed grade 3 subglottic stenosis which was diagnosed on FOB evaluation on previous admission.

Informed Consent was Obtained from the Parents

On arrival to the operating room, standard monitoring i.e., pulse-oximetry, electrocardiography and non invasive blood pressure recording was started. Anaesthesia was

induced with tidal breathing of sevoflurane 2–8% with 100% oxygen. An appropriate sized oropharyngeal airway was inserted and bag mask ventilation was confirmed. Intravenous access was secured with 24 G cannula after achieving adequate depth of anaesthesia. In view of the anticipated difficult airway, difficult airway cart was kept ready with paediatric fibreoptic bronchoscope and other essential armamentarium available.

Tracheostomy being a short procedure was performed under O_2, graded sevoflurane, inj fentanyl with maintenance of spontaneous respiration. Intra op period was uneventful. Procedure was successfully completed. The child was shifted to PACU with spontaneous respiration, hemodynamically stable with 98% saturation on oxygen.

Discussion

Fraser syndrome is a rare, autosomal recessive inherited disorder characterized by presence of primarily cryptophthalmos (Fig. 1) and syndactyly (Fig. 2). This may also be associated with cardiac, laryngeal, tracheal, genitourinary and gastrointestinal abnormalities. 13% of Fraser syndrome children have an associated congenital cardiac defect. ASD, VSD and pulmonary artery anomalies have been reported so a pre-operative echocardiogram is desirable [4].

The diagnostic criteria for Fraser syndrome are divided into major criteria (cryptophthalmos, syndactyly, ambiguous genitalia, urinary tract abnormalities, positive family history) and minor criteria (kidney agenesis, umbilical hernia, congenital malformations of the nose, ear and larynx, cleft lip and palate, skeletal defects and mental retardation). The diagnosis is confirmed by the presence of two major criteria and one minor criterion or one major and at least four minor criteria. Prenatal diagnosis is possible and desirable [5].

Fig. 1 Cryptophthalmos

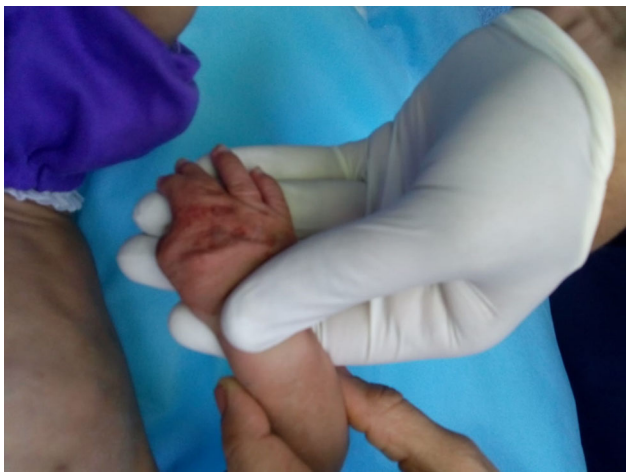

Fig. 2 Syndactyly

Amongst the above, airway anomalies are of paramount importance to the anaesthesiologist. Abnormal airway spectrum includes high arched palate, laryngeal atresia and hypoplasia, laryngeal stenosis etc. Amongst all, subglottic stenosis is the most frequent laryngeal anomaly (83%). Airway abnormalities cause significant problems with management and most of these children require tracheostomy early in life [6]. Laryngeal intubation difficulties and inability to intubate has often been reported in literature [4, 6, 7]. Subglottic stenosis without clinical signs has also been described [8].

Selection of the modality to safely secure airway should be done judiciously. The various choices available can be awake intubation, either blind or with direct laryngoscopy, gum elastic bougies, fibreoptic bronchoscope or an illuminating intubating stylet etc. [9].

In our case, tracheostomy was planned as the airway had been instrumented multiple times. The infant had a risk of developing life-threatening glottic oedema if a definitive airway was not secured. Mathers et al in a review reported a similar case where despite successful mask ventilation it was impossible to intubate the trachea orally even with the smallest ETT available and subsequently required an emergency tracheostomy [10].

Anaesthesiologist should carefully assess the airway prior to anaesthetising these children and a constellation of difficult airway equipment made readily available. An ENT

surgeon should be engaged before induction of anaesthesia in anticipation of airway compromise.

Prompt recognition, adequate preparation and timely involvement of skilled personnel are key to successful management of children with Fraser syndrome.

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Compliance with Ethical Standards

Conflict of interest :The authors declare that they have no conflict of interest.

Informed Consent Informed consent was obtained from the parents.

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