

ANAESTHETIC MANAGEMENT IN A CASE OF OPITZ –FRIAS SYNDROME : A CASE REPORT.

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Background: Opitz-Frias syndrome also known as hypertelorism-hypospadias syndrome or G-syndrome was first reported by Opitz & co-workers in 1969. Syndrome is inherited as X-linked or autosomal dominant trait with male sex limitation. This is a developmental defect with multiple malformations. Interestingly, the birth defects are the defects of midline structures, i.e., telecanthus, hypospadias, congenital heart defects, laryngotracheoesophageal (LTE) defects, cleft lip and palate etc.

Case-report: We report a successful anaesthetic management in a case of Opitz-oculo-genito-laryngeal syndrome in a 5 yr. old male child presenting with hypospadias. This patient had mild expression of the characteristic morphologic abnormalities and possible neuromuscular dysfunction of the oesophagus creating feeding problems and aspiration

Conclusion: Although rare, whenever the diagnosis of this syndrome is suspected as suggested by characteristic facies and male genital abnormalities; the severity of associated anomalies should initiate a thorough multisystem evaluation. This would help in appropriate anaesthetic management.

Keywords: anaesthetic management, Opitz syndrome, aspiration.

INTRODUCTION

Opitz and co-workers first described Opitz-Frias syndrome also known as opitz-oculo-genito-laryngeal syndrome in 1969. Since then only about 40 cases have been described till 1984. This may represent an underestimation because several of these patients present to urologists or paediatric surgeons and characteristic facial features may go unrecognized. The exact overall incidence of the condition is not known. Clinical presentation of this syndrome includes hypertelorism, hypospadias and dysphagia in all cases. The syndrome may present as a mild or severe expression of the characteristic morphologic abnormalities. We present anaesthetic management in a case of Opitz syndrome presenting for hypospadias correction.

CASE REPORT

A 5 year old boy presented to our PAC clinic with the presenting complaint of misdirected urinary stream since birth. There was associated history of recurrent choking episodes especially on swallowing liquids, at times leading to cough and difficulty in breathing. There was no history of cyanotic spells, syncope, convulsions, drug allergy or any previous anaesthetic experience. He was second in birth order and born to non-consanguineous parents after an uneventful pregnancy. The sibling of the child had no abnormalities.

Physical examination revealed a distinctive facies with ocular hypertelorism, an upward slant of palpebral fissures, epicanthic folds and broad nasal bridge. His ears were low set with posteriorly rotated pinnae (fig.1). In addition he had brachycephaly, supernumerary teeth and a small palatal sinus in the midline. Examination of the genitalia revealed distal penile hypospadias with chordee, undescended testes in the inguinal area, and an underdeveloped scrotum.

Anthropometric parameters were as follows: weight = 12.4 kg., height = 95 cms, and head circumference = 47.5 cms. Vital signs were normal. Chest examination was normal except for mild pectus carinatum. Cardiac and other system examination was normal.

Routine investigations, chest X ray and EKG were within normal limits. Indirect laryngoscopic examination could not be carried out as the child was uncooperative. He was accepted for surgery.

Preop preparation

Feeding was withheld 12 hours prior to surgery to avoid choking and aspiration. An IV infusion of paediatric fluid was started at 50 ml./hr., 5 hours prior to surgery to prevent dehydration and hypoglycemia. For aspiration prophylaxis, tablet ranitidine 40 mg. HS and at 6 a.m. was administered in addition to syrup sodium citrate 10 ml. at 6 a.m. Metoclopramide was added 20 min. before surgery. Patient was wheeled in as the first case in the morning. Monitoring of ECG, Oxygen saturation, H. R., B. P. was started. Preoperative vitals were as follows: P. R.: 90/min., B. P.: 106/60 mm Hg. SpO₂ = 100% on room air. ECG showed normal sinus rhythm. Patient was premedicated with 0.2 mg. atropine intravenously. E.N.T. surgeons were called for and were present in the O. T.

Induction was carried out with morphine 1.5 mg. and thiopentone sodium 50 mg. IV in the slight anti-trendelenberg position. Cricoid pressure was applied after the patient lost his consciousness. After ensuring unobstructed ventilation with bag and mask, with 100% O₂ using J. R. circuit; airway was maintained. Neuromuscular blockade was achieved with succinylcholine 25 mg. before laryngoscopy. Laryngoscopy was carried out using straight blade. Vocal cords and larynx appeared normal. Trachea was intubated with 4.5 mm. ID uncuffed PVC tube. After securing tube at 15 cms at the angle of mouth, oropharyngeal packing was done with saline soaked roller gauze. Balanced anaesthesia was maintained with 1-2 MAC halothane with O₂ and N₂O in a ratio of 1:1 and vecuronium, 1.5 mg bolus followed by intermittent dosages of 0.25 mg IV. Analgesia was obtained by giving caudal block with 6 ml of 0.25% bupivacaine.

Vital signs remained within 90% of pre-induction values during the perioperative period. Towards the end of surgery, ryle's tube was inserted and removed after thorough suctioning, residual neuromuscular blockade was reversed using neostigmine and atropine. Oropharyngeal suctioning was done and trachea was extubated in head down lateral position, after ascertaining adequate tidal exchange. Child was fully awake, responding to verbal commands.

Post-operative course

In the immediate post-operative period patient was found to have mild inspiratory stridor along with persistent cough and hoarseness. Forward jaw thrust was applied and gentle positive pressure ventilation given. Inj. hydrocortisone 50 mg intravenously was given along with ondansetron 2 mg.i.v. Stridor subsided in 10 min. Patient was shifted to recovery room for observation and further management. In the recovery room, patient was put on humidified oxygen enriched air via venturi-mask with $FiO_2 = 0.4$. Patient was nursed in the head-up position and was comfortable and pain-free. Parents were advised to keep the patient fasting for further 24 hours and intravenous fluid continued till then. Oral suctioning was advised as and when required. Patient was discharged on the seventh post-operative day in a satisfactory condition.

DISCUSSION

Inheritance: It appears that the syndrome is transmitted as an X-linked or autosomal dominant trait with male preponderance. In the majority, the syndrome was transmitted by mothers with telecanthus to their male offspring. The great majority of affected patients have been males, although two fatal cases in females have been diagnosed.

Features: The syndrome is characterised by functional, craniofacial and genital abnormalities. It is interesting to note that the birth defects seen in the hypertelorism-hypospadias syndrome are the defects of the midline structures.

The **craniofacial abnormalities** are mild to moderate hypertelorism, slight slanting of palpebral fissures, epicanthal folds, an unusual prominence of parietal and occipital regions with associated micrognathia and high arched palate.

The **functional abnormalities** are dysphagia associated with recurrent aspiration, achalasia of the oesophagus (neuromuscular dysfunction of oesophagus) and stridulous respiration. These are reflected in intermittent pulmonary difficulties with associated wheezing and clinically a weak hoarse cry.

The **genital abnormalities** are hypospadias associated with a bifid scrotum.

Less common associations, presenting only in those with the more severe manifestations of the syndrome include cleft lip and palate, short frenulum of the tongue, laryngotracheal cleft (or malformation of the larynx), unusually high carina and hypoplasia of both the pulmonary and vascular components of the lungs.

A child with Opitz-Frias syndrome may present in early life to the anaesthetist with a variety of problems. The greatest threat to life for these children is aspiration. Whenever the diagnosis of Opitz syndrome is suspected in a newborn male, all circumstances that might lead to aspiration must be avoided.

Anaesthetic considerations

- The features of small mouth relative to age and head size, a relatively large tongue, a prominent occiput, a small larynx and small post nasal space contribute to difficulty in intubation in these children.
- Incompetence of the lower oesophageal sphincter increases the likelihood of regurgitation and is an added risk factor.
- Laryngeal hypoplasia is also well documented in these cases as is LTE cleft.

GOALS

- ✓ PAC assessment should lay an emphasis on airways and other system involvement.
- ✓ Preoperative sedation should be avoided.
- ✓ Administration of particulate antacids is not considered appropriate.
- ✓ Facilities to intubate the trachea and to perform an immediate tracheostomy should be available.
- ✓ A full range of tracheal tubes should be kept in readiness.
- ✓ Correct positioning of the patient and adequate amount of neuromuscular blockade is important in maintaining the airway and at laryngoscopy.
- ✓ The possibility of regurgitation and lung soiling should be reduced by keeping the patient adequately fasting, aspiration prophylaxis, a well fitting endotracheal tube, and application of cricoid pressure.
- ✓ Evacuation of gastric contents by nasogastric tube is an effective measure.
- ✓ Because endotracheal intubation may be difficult or impossible; until ventilation is known to be feasible, muscle relaxants should not be administered because successful pulmonary ventilation may not be possible using a face mask and oral airway.
- ✓ In addition to routine monitoring of H.R., B.P., temperature, ABGs, respiratory exchange should be continuously assessed.
- ✓ Danger of aspiration should be kept minimum and patient should receive supplementary oxygen in the postoperative period.

Thus, with the aid of simple and adequate precautions, potential hazards of anaesthesia in a child with Opitz syndrome can be reduced.

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Figure – 1

