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Case Report Of Pierre Robin Sequence With Severe Upper Airway Obstruction Rescued By Tracheostomy

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Abstract

The Pierre Robin sequence (PRS) is classically described as a triad of micrognathia, glossoptosis, and airway obstruction. Infants frequently present at birth with a hypoplastic mandible and difficulty breathing. The smaller mandible displaces the tongue posteriorly, which results in obstruction of the airway. Typically, a wide U-shaped cleft palate is also associated with this phenomenon. PRS is not a syndrome in itself, but rather a sequence of disorders, with one abnormality resulting in the next (1) .Cases with severe respiratory distress due to upper airway obstruction immediately after birth are very rare. (2)

Here we report a case of a newborn who had micrognathia, cleft palate and glossoptosis and was diagnosed as Pierre Robin sequence. In this case tongue protruded into the nasal cavity via a cleft palate occupied pharynx and nasal cavity, resulting in severe respiratory distress. Oropharyngeal intubation was difficult to do. Therefore, an emergency tracheostomy was done to secure the airway for resuscitation. No family history of any congenital disorders was found. Various referrals were made, including to an ear, nose and throat specialist for the management of a tracheostomy tube, paediatric surgery in view of cleft palate and pneumothorax, along with Plastic and reconstructive surgery in view of tongue to lip adhesion surgery, Physiotherapy and paediatric cardiologist. The plan includes trials of complete tracheostomy closure and decannulation. And if the saturation is not maintained then plan to do jaw lengthening procedures (Mandibular distraction osteogenesis)

Keywords: Pierre Robin sequence, Micrognathia, Glossoptosis, Airway obstruction, Cleft palate, tracheostomy, tongue to lip adhesion surgery

Introduction

PRS is characterised by a classic triad of micrognathia, glossoptosis, and airway obstruction which occurs in 1/8500 to 1/14,000 live births and is often accompanied by cleft palate(3,4) Cases with severe dyspnea due to upper airway obstruction immediately after birth are very rare. PRS often develops upper airway obstruction or feeding difficulty secondary to micrognathia, glossoptosis, or a shifted tongue that comes in contact with the pharyngeal wall.(5) PRS is not a syndrome in itself, but rather a sequence of disorders, with one abnormality resulting in the next.The pathophysiological origin of the embryonic sequence of events leading to the disorder seems heterogeneous(6,7) A family history of PRS is present in 10% to 15% of cases, possibly involving mutations upstream of the SOX9 gene, which participates in embryonic mandible development among other roles(8,9) Neonates with PRS exhibit breathing and feeding issues, including upper airway obstruction, sucking and swallowing difficulties and gastroesophageal reflux(10,11).Because of these functional disorders, the first year for an infant with PRS is marked by many burdensome medical necessities such as prolonged hospitalisation,

mother/child separation, nasogastric or gastrostomy tube feeding and management of upper airway obstructions .

Case Report

We report a newborn baby boy weighing in at 2700 gm (appropriate to gestational age) born to G3P1L1A1 mother at 38 weeks of gestational age by LSCS with no family history of any congenital anomalies and with normal anomaly scan. Baby didn't cry immediately after birth and required IPPV for 1minute and was admitted in NICU of D.Y. Patil Hospital in view of perinatal asphyxia and respiratory distress.

On admission to NICU, Baby was put on Bubble CPAP in view of respiratory distress with Downe's score of 4. Routine investigations with BACTEC were sent and started Inj. CEFTAZIDIME and AUGMENTIN. On respiratory system examination suprasternal, intercostal, substernal and subcostal retractions were seen and respiratory distress increased. Hence ABG was done which suggestive of severe metabolic acidosis. Hence Inj. Noradrenaline and Dobutamine was started. Respiratory distress was not settled and we planned for intubation which was difficult. On careful examination of upper airways, we observed that the baby had U shaped posterior cleft palate, micrognathia and glossoptosis and was diagnosed as PIERRE ROBIN SEQUENCE. Meanwhile the baby had an episode of desaturation and bradycardia which required resuscitation. Hence CPCR was started and the baby was revived. Endotracheal intubation was difficult. Hence emergency tracheostomy procedure was done by the ENT surgeon and was put on ventilator support. On CNS examination neonatal reflexes were sluggish and had one episode of GTCS convulsion for which phenobarbitone was started. Respiratory distress was not settled even after tracheostomy, chest x ray was done which was suggestive of right sided Pneumothorax for which ICD insertion was done. On cardiovascular examination no murmur was appreciated and 2D echo suggestive of good biventricular function and severe PAH with PFO left to right shunt and inotropes were continued. Repeat 2D echo was done after 4 days which was normal. Hence inotropes were tapered and stopped. Sepsis screening was done and blood culture was repeated which was positive. Hence escalation of antibiotics therapy was done.

Patient was on a ventilator with minimal settings. As respiratory distress settled and baby was hemodynamically stable plan was to do video laryngoscopy. On Day 10 of life, Diagnostic video laryngoscopy was done where epiglottis, bilateral Arytenoids were visualised with absence of laryngeal cleft and the vocal cords could not be visualised, anteriorly placed larynx. Gradually ventilator settings were tapered and off ventilator trials were given and the baby was maintaining saturation on room air with no signs of respiratory distress. We started with tube feeding which was tolerated and then Wati spoon feeds trials were given and the baby tolerated the feeds well with adequate weight gain. The Cuffed Tracheostomy tube was changed every 16 days. For further management on the 40Th day, the baby was shifted to a higher Centre where tongue to lip adhesion surgery was done and trial of (50%) closure of a tracheostomy tube was done. Currently the plan includes trials of complete tracheostomy closure and decannulation.

Plan of action: to do jaw lengthening procedures (Mandibular distraction osteogenesis) if the saturation is not maintained.

Volume 7, Issue 5; September-October 2024; Page No 276-280

a)Right and Left profile pictures – Micrognathia

B) Posterior U shaped Cleft palate

Discussion

We reported one PRS case that developed severe upper airway obstruction immediately after birth and were rescued by tracheostomy. PRS refers to the association of micrognathia and glossoptosis and palatal malformation is characterised by varying degrees of upper airway obstruction. PRS may occur alone or in association with other syndromes such as stickler syndrome, velocardiofacial syndrome. In about 30% of cases PRS may be an isolated occurrence, while in the following 30% it is related to other anomalies and in the last third of cases it is part of a more complex syndrome i.e.most frequently Stickler Syndrome (12).In our case no association with other anomalies were observed. Feeding difficulties are common as infants struggle to breathe during eating. Gastroesophageal reflux and aspiration are common sequelae of this process.

The associated cleft prevents the formation of negative intraoral pressure, which is required to suck milk from

Volume 7, Issue 5; September-October 2024; Page No 276-280

the breast or bottle; the micrognathia and glossoptosis further impede mechanical sucking(13) Airway obstruction and resulting negative intrathoracic pressures have been identified as factors associated with increased gastroesophageal reflux.(14) Given the poor caloric intake associated with reflux and difficulty feeding and the increased respiratory effort driving increased energy expenditure, these infants often fail to thrive and are unable to gain weight during the early postnatal period. If left untreated, prolonged airway obstruction can lead to acute or chronic hypoxia, cyanosis, apnea episodes, aspiration, respiratory tract infection. Subsequent complications of chronic hypoxia are chronic carbon dioxide retention,elevated pulmonary vascular resistance, cor pulmonale, right heart failure and cerebral hypoxia(15,16)

Most of the patients with PRS get relieved by conservative measures, however patients with pronounced micrognathia, failure to thrive, prolonged use of nasopharyngeal airway, tracheostomy and prolonged endotracheal intubation needs surgical correction. As in our case conservative measures along with surgical approach were there to manage the symptoms of neonate.

The case highlights a multidisciplinary team to assess the anatomic findings, delineate the source of airway obstruction, and address airway and feeding issues encompassing medical, and therapeutic interventions to improve his overall quality of life. Close monitoring and adjustments to the treatment plan are essential for his ongoing well-being.

Conclusion

It is our responsibility as physicians to recognize this disorder timely by proper clinical examination soon after birth, to provide close follow-up and appropriate therapy and counselling. Delay in treatment of the Pierre Robin syndrome is so dangerous that it is essential that all babies with cleft palate should be referred immediately after birth. Prognosis of a PRS is poor as each and every case of PRS is unique and needs to be assessed individually. They are more prone to hypoxia which can lead to neurological deficits. Financially also it is difficult for the parents to take care of the baby at home and to deal with complications like aspiration pneumonia. Antenatal scans should be more effective and efficient so that PRS can be easily detected with accuracy. All the cases of PRS should be thoroughly investigated to diagnose association with other syndromes and to formulate the further line of management.

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Volume 7, Issue 5; September-October 2024; Page No 276-280

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Volume 7, Issue 5; September-October 2024; Page No 276-280