ORIGINAL RESEARCH PAPER

Paediatrics

KEY WORDS:

A CASE REPORT OF PIERRE ROBIN SYNDROM

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INTRODUCTION

Pierre Robin syndrom is characterized by the clinical triad of micrognathia (mandibular hypoplasia), glossoptosis (downward displacement of the tongue), and upper airway obstruction. Clinically, this results in a small, underdeveloped mandible that causes the base of the tongue to fall back into the throat and can ultimately lead to upper airway compromise. It is commonly associated with cleft palate. The sequence was first described in 1891 by Pierre Robin. It affects approximately 1 in 8,500 to 1 in 14,000 newborns in a year.

Etiology

The etiology of pierre robin syndrome is typically divided into isolated(Non Syndromic) and Syndromic. Non-syndromic pierre robin syndrome has been associated with mutations on chromosomes 2, 4, 11, or 17. Some evidence suggests SOX9 or KCNJ2 mutations (on chromosome 17) may affect the development of facial structures and cartilage development, leading to this. Syndromic pierre robin syndrome has associated with Stickler syndrome, Velocardiofacial syndrome, Treacher Collins syndrome & many more.

Pathophysiology

Pierre Robin syndrome is a sequence because one initial malformation leads to a sequential chain of events causing the other anomalies. In Pierre Robin syndrome micrognathia is the first abnormality that leads to glossoptosis and, ultimately, airway obstruction and/or a cleft palate. If mandibular growth is abnormal, the tongue is not able to follow the normal growth and blocks the closure of the palatal cleft. The glossoptosis, due to abnormal mandibular growth, ultimately leads to upper airway obstruction. In some cases like multifetal gestation, structural uterine anomalies, or oligohydramnios the external forces cause the fetal head to become flexed, compressing the mandible against the chest, mandible unable to grow appropriately. The tongue continues to grow normally and is ultimately displaced backward, causing potential obstruction of the upper airway. The tip of the tongue may also impede the fusion of the palatal shelves, leading to cleft palate.

Case Presentation

A pre-term female neonate born at 36 weeks of gestation by normal vaginal delivery out of non consanguineous marriage with birth weight 2.2 kg , appropriate for gestational age, cried immediately after birth to 26 years old 3rd gravida mother. On examination patient have micrognathia, glossoptosis cleft palate with respiratory distress.

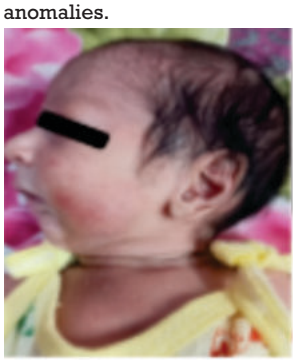
Treatment

Mild disease can often be treated using conservative management without surgery. Baby put on prone and lateral position to allow gravity to pull the tongue anteriorly and improve airway obstruction. Continuous positive airway pressure (CPAP) is a useful intervention that has shown great benefit, but compliance is very difficult. More severe cases require surgical management. Surgical options include tongue-lip adhesion, mandibular distraction osteogenesis and tracheostomy.

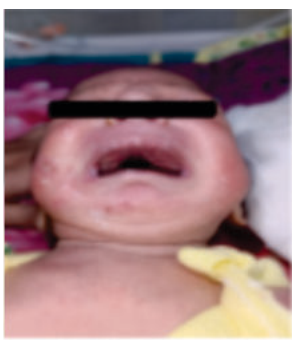
(1) Tongue-lip adhesion is a procedure where the tongue is sutured to the mucous membrane and muscle of the lower lip to hold the tongue in an anterior position in an attempt to reduce the amount of airway obstruction.
(2) Mandibular distraction osteogenesis is surgical intervention that produces long term results. This procedure advances and elongates the jaw by osteotomy follow by new bone formation.
(3) Tracheostomy is a procedure where the trachea is directly cannulated through the anterior aspect of the neck. This is more likely to be performed on patients with airway obstruction in multiple areas. This remains the gold standard of treatment for airway protection.

COMPLICATION & PROGNOSIS

The complications of Pierre Robin syndrom are due to airway obstruction. Short term complications from airway obstruction include desaturations, difficulty feeding, and aspiration. Long term complications may be attributed to hypoxic injury and inability to feed well. This may include cerebral impairment, pulmonary hypertension, cor pulmonale, and failure to thrive. Obstructive episodes can lead to hypoxemia, hypoventilation, malnutrition, asphyxia, cor pulmonale, or even death. Syndromic Pierre Robin syndrome have higher mortality rate. Mortality was associated with cardiac and central nervous system (CNS) anomalies.



Pic.1 Micrognathia



Pic.2 Cleft Palate

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[1] Nelson essentials of pediatrics 9th international edition , section 9, chapter 50, approach to the dysmorphic child, pp 192.
[2] National center for biotechnology information, National library of medicine,pierre robin syndrome.