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Research Article

Pierre Robin Sequence in a Neonate: A Case Report

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Abstract – This paper presents a case of Pierre Robin Sequence (PRS), perinatal asphyxia, and severe respiratory distress in a neonate born at 37 weeks of gestation. The incidence of PRS is reported as 1 in 8,500 in the literature. PRS is a congenital anomaly characterized by mandibular hypoplasia, glossoptosis, and cleft palate. This study discusses the clinical findings of the case and reviews the current literature on PRS.

Keywords – Pierre Robin Sequence, Perinatal Asphyxia, Neonatal Respiratory Distress, Mandibular Hypoplasia, Glossoptosis.

I. INTRODUCTION

Pierre Robin Sequence (PRS) is a congenital anomaly characterized by mandibular hypoplasia, glossoptosis, and cleft palate. Mandibular hypoplasia can lead to posterior displacement of the tongue, causing airway obstruction that requires immediate intervention. PRS is linked to developmental issues of the mandible and may occur in association with syndromes such as Trisomy 18, fetal alcohol syndrome, fetal hydantoin syndrome, velocardiofacial syndrome, or Stickler syndrome, or it may present in isolation. The reported incidence of PRS is approximately 1 in 8,500 births [1].

PRS can cause neonatal respiratory and feeding difficulties and, in older children, may lead to growth retardation and cor pulmonale due to chronic hypoxia. This paper discusses the clinical features of a neonate with PRS born at 37 weeks of gestation with severe respiratory distress, and reviews current therapeutic approaches based on recent literature.

II. CASE PRESENTATION

A 24-year-old healthy father and a 20-year-old healthy mother, third-degree cousins, delivered their second child at 37 weeks and 4 days of gestation via cesarean section. The neonate weighed 2,130 grams at birth, with a nuchal cord present. The infant exhibited no spontaneous respiratory effort, cyanotic skin, a heart rate <100 bpm, meconium staining, and severe mandibular hypoplasia. Neonatal resuscitation, including placement of a laryngeal mask airway (LMA), was initiated in the delivery room. The neonate

was transported to the neonatal intensive care unit (NICU), where the LMA was replaced with an endotracheal tube (ETT).

The neonate was diagnosed with perinatal asphyxia, and passive hypothermia therapy was initiated. Due to vascular access difficulties, a central umbilical venous catheter was placed. An orogastric tube was inserted, and arterial blood gases and chest X-rays were obtained, showing findings consistent with neonatal pneumonia and transient tachypnea of the newborn (TTN). Empirical antibiotic therapy with ampicillin and gentamicin was started, and maintenance fluids were administered according to postnatal age.

Despite optimal mechanical ventilation, the infant's oxygen saturation remained between 75% and 80%. High preductal-postductal oxygen saturation differences, diminished femoral pulses, and a systolic murmur (grade 2-3/6) raised suspicion of ductus-dependent congenital cyanotic heart disease. Alprostadil was administered to maintain ductal patency, and inotropic support was provided for hypotension. Cranial ultrasonography revealed an anterior-posterior diameter of 13 mm in the right lateral ventricle and linear echogenic septations within bilateral lateral ventricles, suggestive of prior hemorrhage.

The clinical course deteriorated despite intensive treatment. The family was informed about the neonate's critical condition and prognosis. Following profound desaturation and bradycardia (heart rate <50 bpm), positive pressure ventilation was administered. Despite adequate ventilation of both lungs, cardiac function did not improve. Cardiopulmonary resuscitation (CPR) was initiated and maintained for approximately 45 minutes, with adrenaline (0.01 mg/kg) administered every 3-5 minutes. The neonate's rhythm remained asystolic, and death was declared.

III. DISCUSSION

Diagnosis of PRS is based on the triad of micrognathia, glossoptosis, and upper airway obstruction. These anomalies occur sequentially, with mandibular hypoplasia leading to glossoptosis, which in turn obstructs the upper airway and prevents proper palatal fusion, resulting in cleft palate in 80-90% of cases. PRS can also be associated with genetic syndromes such as Stickler, Treacher-Collins, and 22q11.2 deletion syndromes [2].

Genetic testing and antenatal diagnosis are pivotal for managing PRS. Accurate genetic diagnosis facilitates targeted treatment plans and counseling regarding recurrence risks in future pregnancies [3].

While prenatal screening for PRS is challenging, certain modalities can assist in early detection:

1. **Ultrasound:** Studies suggest that measurements such as the inferior facial angle, fronto-nasal-mental angle, or facio-mental angle are predictive of PRS. Sensitivity and specificity for these methods range from 84-95% and 81-95%, respectively.
2. **Fetal MRI:** In cases of suspected micrognathia, fetal MRI can assess the severity of glossoptosis and detect isolated cleft palate.
3. **Genetic Testing:** Prenatal genetic testing should be considered in pregnancies where routine screening suggests potential anomalies.
4. **Counseling:** Early diagnosis enables improved understanding of PRS and facilitates specialized care planning.
5. **Delivery Planning:** Neonates diagnosed with PRS should be delivered at tertiary care centers equipped for advanced airway management and NICU support.

Management of PRS requires prompt recognition and intervention for airway obstruction, which is potentially life-threatening [4], [5].

Treatment strategies include both non-surgical and surgical approaches:

Non-Surgical Options:

- Prone positioning
- Nasopharyngeal airways
- Palatal plates (e.g., pre-epiglottic baton plates)
- Continuous or intermittent positive airway pressure (CPAP/IPAP)

Surgical Options:

- Tongue-lip adhesion
- Mandibular traction
- Subperiosteal release of the floor of the mouth
- Mandibular distraction osteogenesis
- Tracheostomy

IV. CONCLUSION

Early and frequent evaluation of upper airway obstruction in PRS is critical for optimal management. Despite various treatment algorithms and classification systems, there is no consensus on best practices. Given the heterogeneous phenotypic presentation of PRS, affected children should receive multimodal evaluation and care at specialized tertiary centers.

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