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Research Article

Neonatal Pierre Robin syndrome (Systematic Review)

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Abstract

The early hour life of newborn is an ambiguous and challenging period in human life which need careful and intelligent monitoring to obtain better life outcomes. Pierre Robin Sequence one of the rare syndrome that is observed shortly after or before the birth. PRS is a group of malformation in the baby that develop during pregnancy. It is called a casade because one of its definite features – an undeveloped lower jaw – initiate a casade of other deformities while the baby is developing in uterus and can ultimately lead to post-natal upper airway obstruction and life threatening condition that lead to eventual death.

Aim: The purpose of the study was to highlight the significance of three-dimensional ultrasound in the screening of fetal micrognathia and highlight the diagnostic features of infant with Pierre Robin syndrome; to establish an early assessment approach for better outcomes of infant life.

Methodology: A systematic literature review depends on collecting data from an evidence based studies. Searches were made of twenty five electronic databases: the Cochrane Oral Health Group's Trials Register, The Cochrane Central Register of Controlled Trials (CENTRAL), EMBASE, PsycINFO, Scopus and Web of science, MEDLINE (PubMed).

Results: There is a strong association between breathing difficulty, feeding difficulty in infant and the small size of mandible which affect on fetus shelf life.

Conclusion: PRS is a rare anomaly so raise the awareness to this syndrome should be increased between different physicians. Prenatal diagnosis is very important step for appropriate treatment decision by parents before delivery and to arrange with a multidisplinary team to assessment the condition postnatal to reduce the short and long term complications and treatments.

Keywords: Birth Defects; Pierre Robin Sequence; Micrognathia; Glossoptosis; Airway Obstruction; Obstructive Sleep Apnea; Feeding Problems

Introduction

Pierre Robin Sequence (PRS) considered a nonspecific anomaly characterized by a triad of micrognathia, glossoptosis, and airway obstruction that may occur either isolated defect or as a broader group of malformations. Infants often present at birth with a hypoplastic mandible (which termed "mandibular hypotrophy") and glossoptosis (an aberrant tongue retraction), this condition of the small mandible displaces the tongue posteriorly, resulting in obstruction of the airway.

Mortality for infants with PRS ranges from 1.7% to 11.3%; the rate increases to 26% when examining only the subset of syndromic patients. Airway obstruction compromises breathing

and oral feeding and later on speech problems, due to the difficulty in glossopharyngeal laryngeal vagal function, so the progression to oral feeding is another marker of improvement the airway obstruction [1,2].

Feeding function is the main target process that the newborn must master. This natural process require a complex coordination of sucking, swallowing and breathing, Thus need effective communication between neurologic, respiratory and gastrointestinal systems. Sucking, is the first fundamental component of feeding which require backward and forward movement of tongue. Swallowing is the next important mechanism of feeding that is explained in four phases: oral preparatory phase when bulous is formed, oral phase: during moving bulous of food toward oropharnyx, pharyngeal phase: during pharyngeal constrictors move food bolus toward osophagus and simultaneous close the larynx and so nasophaynx protected by soft palate, Esophageal phase: osophagus transmit food involuntary to stomach [3].

Birth defects like the anatomic or neurogenic abnormalities can disrupt and negatively impact on feeding process. Discordance manifested as prolonged feeding time, oxygen desaturations during feeding, dysphagia, gastroosophageal reflux and aspiration. Infants struggle to maintain respiratory during feeding may be misconstrued as "fussiness" or unusual sleepy which can delay or mask the necessary interventions needed. Early assessment of feeding is very critical factor in the initial airway assessment of infants with PRS [4].

The incidence of PRS is 1/8500–1/14,000 delivery. There are several reasons explained this condition, the most popular is the mechanical theory. This theory considers the impact of various external factors on the fetus in intrauterine period such as oligohydramnios, breech position, or abnormal uterine anatomy. This external forces cause the fetal head to become flexed, compressing the mandible against the chest, rendering it unable to grow appropriately. The mechanical theory is completed by the neurological theory and the genetic theory. The neurological theory refers to a delay in neurological maturation observed in the electromyography for the tip of the tongue and a delay in hypoglossal nerve conduction. The genetic theory postulates the chromosomal deletions, genetic mutation or genetic error [5,6].

Pierre Robin sequence can be associated with cleft palate. However, cleft palate is not a strict criteria for the diagnosis of PRS. It is also associated to another craniofacial anomalies in conjunction with characteristic different syndromes. Stickler syndrome, an autosomal dominant condition, is characterized by short mandibular ramus, antegonial notching of the mandibular body, myopia, and joint problems. Another form associated with PRS, Velocardiofacial syndrome is characterized by a retrognathic mandible, palatal abnormalities, impaired thymus development, hearing loss, pulmonary atresia, hypothyrodism and cardiac malformations [7]. Diagnosis of PRS or craniofacial defects may start in the prenatal period with ultrasound or MRI.

Prenatal ultrasound examination is critically important in the screening of perinatal birth defects as it is simple, fast, and safe. Micrognathia could cause difficulty breathing and feeding due to severe mandibular retrusion and obstruction of the airway. Micrognathia may be difficult to diagnose via ultrasound. Normalization of the mandibular anteroposterior length by the biparietal skull width creates a jaw index. Airway obstruction is another a prognostic marker for suspect PRS, It is a life-threatening condition for the neonate. Prolonged obstruction results in hypoxia, apnea, respiratory tract infections, aspiration, compromised feeding, and failure to thrive. Chronic hypoxia leads to increased pulmonary vascular resistance, cor pulmonale, heart failure, and cerebral hypoxia. Monitor feeding sessions for any signs of dysphagia, coughing, choking, or feeding refusal, also can aid in early evaluate the condition and so urgent intervention [8,9].

Obstructive sleep apnea (OSA) is a common complications appear to the infants and children with craniofacial malformations. OSA is an episodes of complete collapse of airway or partial collapse of upper airway with associated decrease in oxygen saturation during sleep. A clinical grading system for airway obstruction devised by Cole., *et al*. There are 3 Grades: Grade 1 = no respiratory distress when nursed supine, Grade 2 = intermittent evidence of mild respiratory obstruction when nursed supine but feeding precipitates some respiratory distress and Grade 3 = moderate to severe respiratory distress when nursed supine, unable to feed orally [10].

Management of PRS of newborn must begin with assessing severity of airway obstruction. In most cases, affected neonates may be treated without surgery, conservative approach using prone/lateral positioning and palatal obturator to clear the obstructed airway. Infants cannot easily fed in the prone position, so those that cannot safely or adequately swallow when supine, require tube feeding assistance with continuous positive airway pressure consider an effective non-invasive treatment method used in sever upper airway obstruction in infants to reduce need for surgical approach so Continuous positive airway pressure consider as a mandatory approach alternative to tracheostomy When surgery has been deemed necessary in sever advanced anomalies, Surgical options include tongue-lip adhesion (or glossopexy), mandibular distraction osteogenesis, subperiosteal release of the floor of the mouth, and tracheostomy with additional surgical options used more selectively Tracheostomy consider the first line surgical modality used [11].

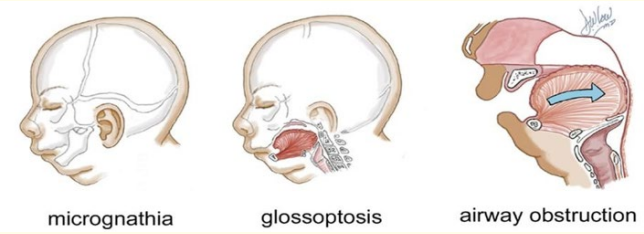

Figure 1: Pierre Robin Sequence triad: micrognathia, glossopteris, upper airway obstruction.

Discussion

PRS is described as a malformation with embryologic dysfunctional pathogenesis, characterise by a classic triad of small jaw, retracted tongue, and air passage obstruction and the anatomic consequent feeding problems. Its etiology that justifying the reason for this syndrome, genetic cause and environmental theories. Infants with PRS should be evaluated by a multidisciplinary team to assess findings, delineate the source of airway obstruction, and address airway and feeding issues. There is no gold standard for evaluation and diagnosis of PRS. During pregnancy, ultrasound findings of micrognathia raise concern for PRS. After birth, an initial evaluation in the delivery room is important to determine if the patient needs immediate airway intervention or no need.

Clinical Evaluation according to the apparent sign and symptoms.

Micrognathia

This is a diagnosis of underdeveloped mandible with a shorter mandibular body length and a greater mandibular angle. However, cannot be helpful in predicting patients who will experience respiratory problems.

Glossoptosis

This is displacement of the base of the tongue towards the pharynx. There is a wide degree of severity related to respiratory distress. There is no standard for diagnosing glossoptosis, but endoscopy and computed tomography (CT) imaging may be useful in quantifying the level of obstruction present.

Airway obstruction

Clinical signs of airway obstruction include abnormal breathing sounds, increased respiratory accessory muscle use, desaturations, difficulty feeding/swallowing, reflux, and aspiration. Long term signs of airway obstruction may include reduced weight gain, difficulty speaking, neurological deficits, and ultimately pulmonary hypertension and cor pulmonale. The clinical severity of airway obstruction, ranging from mild dysfunction not requiring any intervention to severe ventilatory compromise requiring intubation immediately after birth. Episodes of air way Obstruction can progress to hypoxeia, hypoventilation, malnutrition, asphyxia, or even death. Patients with PRS have a higher mortality rate and so show poor prognosis. Mortality was associated with cardiac anomalies. PRS have been classified into groups or grades based on severity. The most common classification system was developed by Cole., *et al*. and reports grades 1-3 from mild to severe. A new classification system by Li., *et al*. describes a 4 group system to assist in the choice of intervention. This system includes grade 0, a very mild form without respiratory or feeding dysfunction [12]. During early postnatal period, these infants frequently fail to live and gain weight due to undernourishment related to reflux and eating issues and high-energy expenditure related to high respiratory exertion. Mild disease can often be treated using conservative management without surgery. Non-surgical options include prone sleeping position, This prone the lateral positioning to allow gravity to pull

the tongue anteriorly and improve airway obstruction, resolving approximately 70% of cases of PRS. If positioning alone is not successful, adjunctive measures such as supplemental oxygen with watchful waiting and follow up the improvement in breathing and feeding. Moreover, a modified nasopharyngeal tubes can be used to bypass the tongue base obstruction. This Non-invasive ventilation by using supplemental oxygen, use of a oropharyngeal or nasopharyngeal tube, and endotracheal tube placement used as a temporary measure to keep the airway open, though special attention must be paid by parents for avoid the complications such as aspiration and obstruction of the tube. More severe cases of PRS require surgical management. Surgical options include tongue-lip adhesion, mandibular distraction osteogenesis, and tracheostomy [13]. Another key feature of our treatment focuses on feeding and nutritional support. All patients are evaluated closely in regards to their ability to feed by qualified speech therapists who specialize in feeding. If necessary, early nasogastric feeding is initiated to supplement oral feeds and improve weight gain. Prior to any surgical intervention, we confirm the absence of obstruction below the level of the tongue base with direct laryngoscopy and bronchoscopy performed by a pediatric otolaryngologist. Tracheostomy remains the gold standard for definitive airway protection.

Conclusion

Pierre robin casade is a rare complex syndrome that is begin in intrauterine life with sever complex outcomes postnatal. Prenatal diagnosis is important for appropriate counseling of parents before delivery for predicte the short and long term complications and treatments for PRS. This also allows all team members to be prepared for intervention at delivery if needed, such as maternal-fetal medicine providers, neonatologists, pediatric anesthesia, maxillofacial surgeon and/or pediatric otolaryngologists. Postnatal assessment for air way obstruction is the first step to evaluate the condition for the intervention needed. Oral feeding is important prognostic marker of improvement the air way condition. A multidisciplinary team approach is important early to address any feeding or airway issues. Doctors must raise the awareness of parents regarding how to deal in this situation to avoid serious unpredicted outcomes.

Fund

Nil.

Conflict of Interest

No conflict.

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