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Case Report

Palatoplasty in a Severe Retrognathic Mandible: A Case Report

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ABSTRACT:
The most common birth defect is the presence of a cleft and 27-34.3% of the clefts in the face are those found only in the palate. The prevalence of ICP varies from 0.17 to 0.7 out of 1000 live births. Females are equally or more affected by a ratio of 1.5-1.6/1 than males. Pierre Robin sequence is a congenital anomaly characterized by micrognathia, cleft palate and glossoptosis at varying levels and presentations. It could be seen as an isolated anomaly or be associated with other congenital conditions or one feature of many syndromes. Several methods have been described in the management of PRS. The main aim of treatment focused at respiratory distress and nutrition. Extreme vigilance is required during every stage of the management of anomalies presented with PRS. Efficient palatoplasty is required to treat the cleft in palatal region so that patient regains the level of proper nutrition and speech in a smooth and speedily manner.
Keywords: Palatoplasty, Pierre Robin sequence, retrognathic mandible.

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INTRODUCTION

A cleft only in the palate is associated with a syndrome in 21-33% of the cases. Cleft palate seems to be caused by a disorder in neural crest migration to the maxillary and palatal fields. The last decades more than 30 genes are associated with cleft palate and presently examined as possible contributors: MSX1, NECTIN1, IRF6, TP63, LOXL3, TBX22, SPECC1L, PHF8, SOX9, TGDS, CDH1, TBX1, SF3B4, SLC17A8, SLC6A9, SLC6A3, BMP4, GABRB3, PRSS12, TAS2R16, FGF8, ACTL6A, ALX1, ALX3, MTHFR, FREM1, FGFR1, ALX4 genes and Chromosomes 2 and 22. Many syndromes may be present together with a cleft in the palate (Table 1). The most common ones are: Stickler syndrome, 22q11.2 deletion (DiGeorge syndrome, velocardiofacial syndrome), Facio-aurico-vertebral/oculo-auricolo-vertebral spectrum, Kabuki syndrome, Treacher Collins syndrome, Fetal alcohol syndrome, Diabetic embryopathy, Down syndrome, Spondyloepiphyseal dysplasia congenital and Kniest dysplasia, and Van der Woude syndrome.[1] Robin sequence is defined as a triad of retrognathia, glossoptosis, and airway obstruction. Prevalence of this disorder ranges from 8,500 to 20,000 births per year.[2] During fetal development, the hypoplastic mandible displaces the tongue into the palatal space, often preventing normal fusion of the palatal shelves.[3] AU-shaped cleft involving the secondary and posterior primary palate is seen in 66% to 90% of affected patients.[4] Some groups erroneously consider cleft palate to be a diagnostic criterion, despite Robin's original definition of the disease.[5] Retropositioning of the tongue base causes airway

obstruction and increased work of breathing. Secondary comorbidities resulting from airway obstruction include failure to thrive, developmental delay, reflux, feeding difficulties, CO2 retention, heart failure, brain damage, and sudden death. The morbidity of upper airway obstruction in an infant can be significant. As Randall described, the infant "can literally exhaust himself to death unless the obstruction is relieved." Upper airway obstruction associated with Robin sequence can be significant, with mortality ranging from 1.7% to 65%. Robin sequence is recognized as a heterogeneous condition caused by isolated mandibular hypoplasia, or by a multitude of conditions that can produce a similar phenotype. More than 50% of affected patients have an associated syndrome, or genetic or medical anomaly, the most common of which is Stickler, although more than 40 syndromes have been associated with Robin sequence. Due to the widely varied nature of presentation, consensus in diagnoses and management remains elusive, producing significant obstacles to formulating definitive treatment protocols.[6] Though in the initial description palatal cleft was not mentioned current literature suggests that micrognathia is the primary anomaly that causes both cleft palate and upper airway obstruction in PRS. Epiglottal hypoplasia though rare has been associated with the condition. Congenital heart disease has been reported in about 20% of PRS patients of which ventricular septal defect (VSD), patent ductus arteriosus (PDA), and atrial septal defect being most common. Recently, a genetic cause to PRS was identified, which may be due to genetic anomalies at chromosomes 2, 11, or 17. A locus for PRS has been mapped to chromosome 17q24 in a four-generation family. Investigators found significantly reduced SOX9 and KCNJ2 mRNA expression in patients, using array comparative genome hybridization analysis with human whole genome oligonucleotide microarray kits 244K in PRS, and the nonsyndromic PRS may be caused by both SOX9 and KCNJ2 dysregulation.[7]

CASE DETAILS

A 1-year-old child patient reported to our clinic with the chief complaint of difficulty in swallowing. On clinical examination, it was observed that after intubation on keeping the mouth gag it found that patient was having less than one finger mouth opening. The patient had cleft of soft palate with severe retrognathia of mandible. The case was diagnosed as of Pierre Robin's syndrome. Under such circumstances, proper palatoplasty was performed with the help of radical muscle. All the investigation reports were well within normal ranges.

DISCUSSION

"Pierre Robin" is a readily recognized condition consisting of a hypoplastic or retrognathic mandible and glossoptosis leading to respiratory distress, with or without a cleft palate which when present is usually a wide "U" shaped cleft, though "v" shaped or soft palate clefts have been reported.

Figure 1- Severe retrognathia of the mandible was evident in the patient

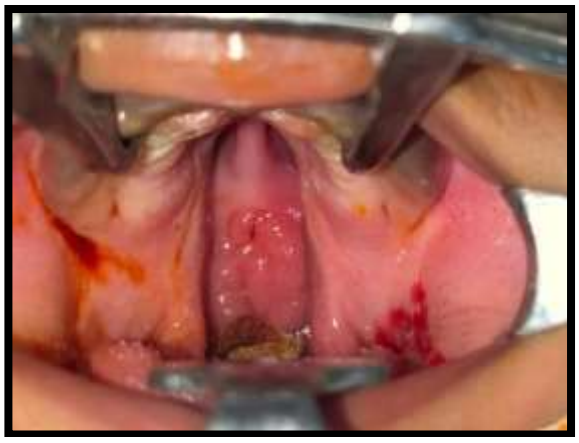

Figure 2- Patient had midline cleft in the soft palatal region

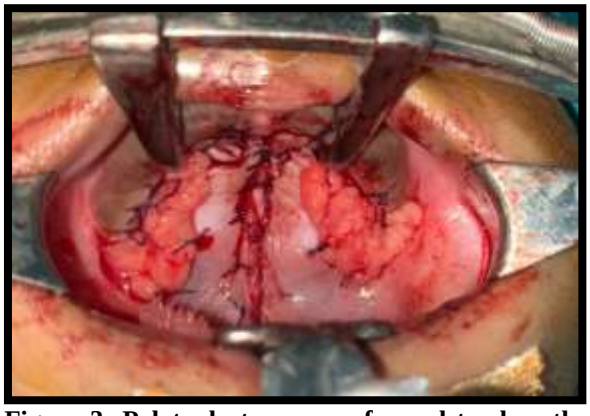

Figure 3- Palatoplasty was performed to close the midline cleft

Though multiple anomalies are present, it is considered as a "sequence" rather than a syndrome as one or all anomalies are possible consequences of the mandibular anomaly. The incidence reported varies

between 1:8,500 and 1:14,000 live births.[8] Management is focused mainly with functional issues associated with airway and feeding which was also noted in our case where the patient suffered from feeding difficulties. A 30% mortality has been mentioned, but the prognosis is good if the neonate survive's the respiratory and feeding issues. Thus, treatment may begin as a surgical emergency in the neonatal period to the symptomatic management of upper respiratory obstruction and feeding problems. All procedures are mainly directed to widening the pharyngeal space or bridging the narrow upper airway.[9] Vipulananthan et al. have reported that male gender and primary presentation with respiratory failure predicted more severe airway obstruction and the need for tracheostomy in patients with PRS. In their study, patients with PRS with cleft palate more frequently showed early respiratory failure than patients with no cleft.[10] PRS is heterogeneous in terms of both diagnostic criteria and clinical consequences. Predicting the severity of respiratory and digestive disorders in newborn infants with PRS could help to reduce both morbidity and mortality rates and to choose the most suitable treatment. Due to the lack of consensual guidelines for assessing and managing respiratory and feeding disorders in the neonatal period, therapeutic options widely vary among pediatric teams and comparing the results of previous series is a difficult issue. Regarding airway obstruction, conservative treatments include prone position, palate plates, nasopharyngeal tube, and non-invasive ventilation; and surgical treatments include mandibular distraction osteogenesis (MDO), tongue-lip adhesion (TLA), and tracheostomy. Feeding disorders, mainly due to sucking–swallowing and esophageal dysfunctions, may cause failure to thrive and aspiration pneumonia and require enteral feeding using nasogastric tube (NGT) or gastrostomy, in up to 75% of patients with PRS. Even if a direct causal link between anatomic features and functional disorders may be reasonably suspected, this has not been yet demonstrated.[11]

CONCLUSION

It is imperative that the closure of palatal cleft should be performed at 18 months after certain advancement in mandibular growth before beginning of active speech. Further, it gives better patient compliance reduces the need for tracheostomy so that no postoperative airway embarrassment or any problematic event happens with baby compliance.

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