

A Pierre Robin Syndrome with Absent Anterior 2/3 Tongue—A Case Report

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Abstract

The triad of micrognathia, glossoptosis and airway obstruction originally described in 1923 by Pierre Robin, is known as Robin sequence (or Pierre robin sequence “PRS”). PRS is characterized by micrognathia (small and symmetrical receded mandible), glossoptosis (tongue of variable size falls backwards into the post pharyngeal wall), and cleft palate (U or V shaped). We report a case of 2 hours old newborn presented with micrognathia, retrognathia, and glossoptosis and absent anterior two thirds of tongue.

Keywords

Micrognathia, Glossoptosis, Pierre Robin Sequence

1. Introduction

Robin sequence is an etiologically and phenotypically heterogeneous disorder [1]. PRS occurs as an isolated defect, as a part of recognized syndrome, or as a part of complex of multiple congenital anomalies. Diagnosis of a possible syndrome is very often critically important for correct management of a newborn affected with PRS [2] [3]. Isolated PRS is often a deformation resulting from intrauterine forces acting on the mandible, which restrict its growth and impact of the tongue between the palatal shelves. Some deformational cases of PRS have been associated with oligohydramnios. Because micrognathia results from intrauterine molding, mandibular catchup growth is expected after birth once intrauterine forces are removed. The most severe cases of micrognathia are unlikely to be isolated PRS caused by deformation. Therefore, catchup growth is unlikely.

2. Mini Review of Literature

In patients with PRS, 13% - 27.7% of other family members are affected with cleft lip with or without cleft pa-

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late [4] [5]. Jacobsen *et al.* screened 10 unrelated patients affected with PRS for *SOX9* and *KCNJ2* mutations and suggested that nonsyndromic PRS may be caused by both *SOX9* and *KCNJ2* dysregulation. Several lines of evidence for the existence of a 17q24 locus underlying PRS, including linkage analysis results, a clustering of translocation breakpoints 1.06 - 1.23 Mb upstream of *SOX9*, and microdeletions both approximately 1.5 Mb centromeric and approximately 1.5 Mb telomeric of *SOX9*, have been reported by Benko *et al.* [6] [7].

The proportion of cases that are isolated PRS varies in different studies. Hanson and Smith found that 25% of PRS cases had specific syndromes, another 35% had multiple anomalies without a specific recognized syndrome, and only 40% had isolated PRS [8]. Another study found that 74% of cases were isolated PRS [9].

While there is a great variation in severity, PRS is characterized by the following phenotypic features: micrognathia (small and symmetrical receded mandible), glossoptosis (tongue of variable size falls backwards into the post pharyngeal wall), cleft palate (U or V shaped) [10] [11]. Infants with PRS sequence often have airway obstruction, feeding difficulties, and challenges in gaining weight, and they may have associated anomalies, including hypotonia and limb reduction defects [12]. Congenital heart defects are present in up to 25% of the babies with PRS who die in early infancy. Patent ductus arteriosus (PDA) is the most common, followed by atrial septal defects, ventricular septal defects and coarctation of aorta. It has been reported that more than 20% of individuals will have developmental delay or cognitive impairment, and overall morbidity and mortality are higher in syndromic PRS or PRS with associated anomalies compared with isolated PRS [9].

3. Case Report

19 years old primi with no history of pregnancy induced hypertension, diabetes mellitus, premature rupture of membranes, oligohydromnios and polyhydromnios delivered a female baby by LSCS with birth weight of 2.5 kgs with history of birth asphyxia. On examination at 2 hours of age in hospital child is having micrognathia, retrognathia (Figure 1) and difficulty in breathing. On oral examination child is having “U” shaped cleft palate (Figure 2). Child has inspiratory stridor. Child had difficulty in maintaining saturation. We started on nasal prongs but child did not improve. Then we kept the baby in prone position with hood, then saturation maintained but there were repeated attacks of apnoea, so we thought to intubate the baby. But it was very difficult to intubate for us. We called for anesthetists to do the intubation, even for them it was difficult. With the help of otorhinolaryngologist (ENT Surgeon) we did VLS (Video-Laryngoscopy) to see the vocal cords (Figure 3 and Figure 4). On close examination we concluded that infant is having not only posteriorly placed tongue but anterior two thirds of the tongue is not formed (Figure 5). We kept the child on supplemental oxygenation with hood in thermo neutral environment. As the child is not taking full feeds orally, we kept the child on parental I.V. fluids and orogastric feeds. Child gradually maintained oxygen saturation at room air in prone position.

Child was started orogastric expressed breast milk feeds and gradually started taking palady feeds. The whole process took 3 weeks for the baby to stabilize and for discharge. Follow up after 1 week child is taking palady feeding, sleeping comfortably in prone position. Child started gaining weight. These children have feeding difficulties, they should be taken special interest in feeding with palady with calorie enrichment of milk.

PRS with absent anterior 2/3 of the tongue has not been reported till now. Hence we are reporting this case as a rare entity, after obtaining the informed consent from the patient’s family.

4. Discussion

In infants with PRS, the tongue is displaced toward the posterior pharyngeal wall, resulting in obstruction at the level of the epiglottis. The tongue can act as a ball valve, leading to inspiratory obstruction. In addition to micrognathia, other mechanisms may contribute to airway obstruction in individuals with PRS, such as pharyngeal hypotonia and airway inflammation from associated gastro-oesophageal reflux. Patients with PRS may present in the immediate neonatal period with increased inspiratory work of breathing, cyanosis, and apnoea. Obstruction is more common in the supine position [13]-[15]. Chronic obstruction can lead to failure to thrive, carbon dioxide retention, pulmonary hypertension, and eventually right sided heart failure (corpulmonale). Air way obstruction is the main cause of feeding and growth issues in infants with PRS. Feeding problems can also be related to inadequate tongue control or pharyngeal hypotonia and complicated by presence of a cleft palate. Increased energy expenditure due to increased work of breathing may lead to failure to thrive if infant is not receiving adequate calorie intake [16].

The single initiating defect of this disorder may be hypoplasia of the mandibular area before 9 weeks in utero,



Figure 1. Showing micrognathia, retrognathia.



Figure 2. Showing "U" shaped cleft palate.

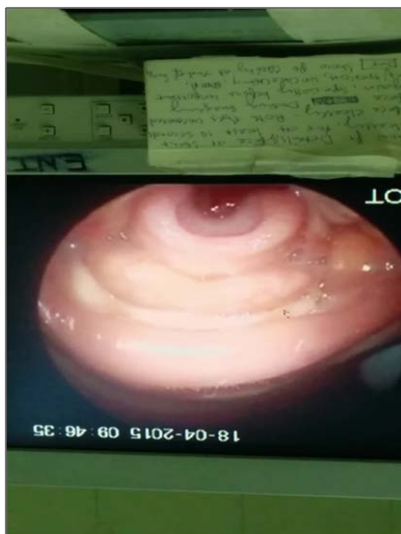


Figure 3. Difficult airway seen with VLS.

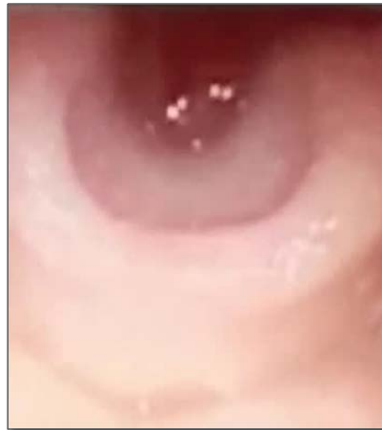


Figure 4. Close view of vocal cords with VLS.



Figure 5. Showing absent anterior 2/3 tongue.

allowing the tongue to be posteriorly located and impairing the closer of the tongue to meet the midline [17] [18] (**Figure 6**) (Smiths text book of recognizable patterns of human malformations, 6th ed., page no. 262).

Anterior two-third of the tongue is formed by fusion of the tuberculum impar and the two lingual swellings. The anterior two-third of tongue is thus derived from the mandibular arch (**Figure 7**) (Inderbeer Singh text book of human embryology, 7th ed., page no. 162). From mandibular arch medial and lateral pterygoids, masseter, temporalis, mylohyoid, anterior belly of diaphragm, tensor tympani, tensor palati are derived [17] [18].

In this case except anterior two-third of tongue all other derivatives of mandibular arch are developed. So when we see an infant is having PRS sequence then we have to check whether the baby is having isolated PRS sequence or syndromic baby having PRS sequence. We must be prepared for the expected complications like difficult airway and tongue abnormalities causing difficult breathing [18] [19].

The tongue in PRS is usually normal in size with foreshortened floor of the mouth and inspiratory obstruction. The infant should be in prone position to relieve inspiratory obstruction. Some patients may need tracheostomy. Mandibular distraction procedures can improve mandibular size. Feeding requires great care, patience and palady feeding.

5. Conclusions

The new born baby presented with micrognathia, retrognathia and “U” shaped cleft palate with breathing difficulty, apnoea and inspiratory stridor. Respiration maintenance was difficult in supine position. Child maintained

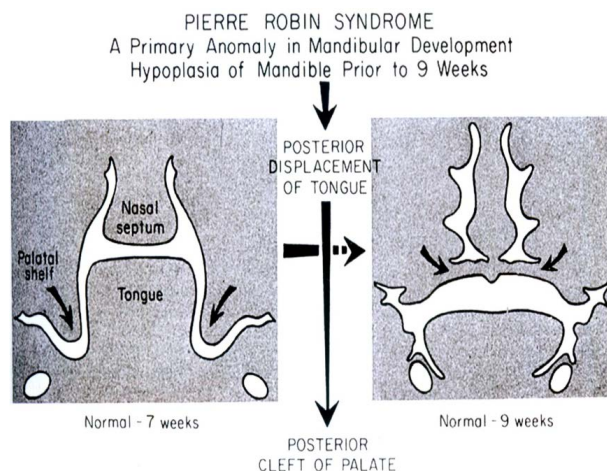


Figure 6. Diagrammatic depiction of development of mandible.

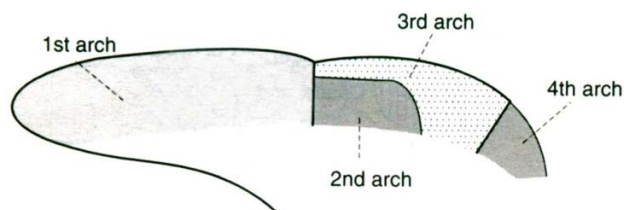


Figure 7. Diagrammatic depiction of development of Tongue.

saturation in prone position. With palady feeding child was improved. Absent anterior two-third of the tongue is the only additional finding, usually not a component of PRS.

For us to observe the morbid conditions like cognitive impairment, developmental delay, growth retardation, we need to follow up the child further.

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