Case Report

Variant of Pierre Robin Sequence Requiring Prolonged Tracheostomy

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Chettinad Health City Medical Journal 2014; 2(2): 63 - 64

Abstract

Pierre Robin sequence (PRS) or anomalad, a well-recognized presentation, is the association of the first branchial arch malformation. It presents with a classic triad of micrognathia, glossoptosis, and cleft palate. Presenting here is a neonate with features of Pierre Robin sequence with syndactyly of fingers and toes and congenital heart disease [ASD with PDA], which also needed tracheostomy on day 16 of life and decannulation done after 7 months.

Key words : Pierre Robin, Tracheostomy

Introduction

Pierre Robin Sequence is considered to be a nonspecific anomalad which may occur either as an isolated defect or as a broader group of malformations[1]. It presents with a classic triad of micrognathia, glossoptosis, and cleft palate. In 1923 French physician Pierre Robin introduced the term 'glossoptosis' in association with micrognathia. He later reported an association with cleft palate in 1934 and this constellation of findings was termed as syndrome[2].

PRS is a clinically well-defined subgroup of the cleft lip-palate population with an unknown etiology, often observed as a part of other Mendelian syndromes, such as Stickler's syndrome, velocardiofacial syndrome, and Marshall's syndrome[3].

Recent studies on genetics have shown that that the association of dysregulation of the genes SOX9 and KCNJ2 may be involved in PRS, evidenced by a familial translocation with a breakpoint located in the gene empty region between SOX9 and KCNJ2, and by reduced expression of SOX9 and KCNJ2 in non-translocated patients with PRS[4].

Case details

Here is a live term male baby second born to a non consanguineously married couple. Mother is an elderly[39 yrs], multigravida with history of three spontaneous abortions. There was no h/o oligohydramnios. Baby was born by lower segment caesarean section indication being previous section, baby cried immediately after birth with birth weight of 2.4kg and on examination had features like low set ears, micrognathia, retrognathia, rudimentary tongue, glossptosis, U shaped cleft palate suggestive of Pierre Robin sequence[Fig 1] and also syndactyly of second and third fingers and toes were noted[Fig2]. On auscultation there was systolic murmur and the baby was managed in the Neonatal intensive care unit. Investigations like ultrasound abdomen and cranium, infantogram done was normal, echocardiography showed ASD [0.6mm] with PDA[0.5mm]. Karyotyping was normal. As baby had respiratory distress and was not maintaining saturation even in prone position required intubation frequently, hence tracheostomy was done on day 16 of life, metabolic parameters were corrected, intravenous antibiotics given and Nasogastric feeds with expressed breast milk started on day 2 of life and baby was discharged on day 36 of life with tracheostomy and nasogastric tube in situ. The mother was trained regarding the maintenance of both the tubes and was called for follow up. She was on follow up, baby was growing well and tracheostomy site was clean, hearing evaluation done at 3 months by otoacoustic emission test was normal, there were no orthopedic problems and at the age of 7 months tracheostomy tube was removed, and now the baby has no respiratory distress and has gained weight accordingly, development is according to age except language milestones where only cooing is present [no monosyllables], no history suggestive of any obstructive sleep apnea and surgery for cleft palate is being planned followed by speech therapy.

Discussion

Pierre Robin Sequence is a congenital abnormality characterized by the presence of a combination of mandibular hypoplasia, glossoptosis and often labio palatine clefting[5].

A relatively small number of patients with clefts of palate are not of the multifactorial inheritance[6]. These patients usually make up approximately 3% of child population and present with one or more additional structural abnormalities[7].

- Three pathophysiological theories exist to explain the occurrence of Pierre Robin sequence[8].

  The mechanical theory: This theory is the most accepted. The initial event, mandibular hypoplasia, occurs between the 7th and 11th week of gestation. This keeps the tongue high in the oral cavity, causing a cleft in the palate by preventing the closure of the palatal shelves. This theory explains the classic inverted U-shaped cleft and the absence of an associated cleft lip. Oligohydramnios could play a role in the etiology since the lack of amniotic fluid could cause deformation of the chin and subsequent impaction of the tongue between the palatal shelves.

- The neurological maturation theory: A delay in neurological maturation has been noted on electromyography of the tongue musculature, the pharyngeal pillars, and the palate, as has a delay in hypoglossal nerve conduction. The spontaneous correction of the majority of cases with age supports this theory.

- The rhombencephalic dysneurulation theory: In this theory, the motor and regulatory organization of the rhombencephalus is related to a major problem of ontogenesis.

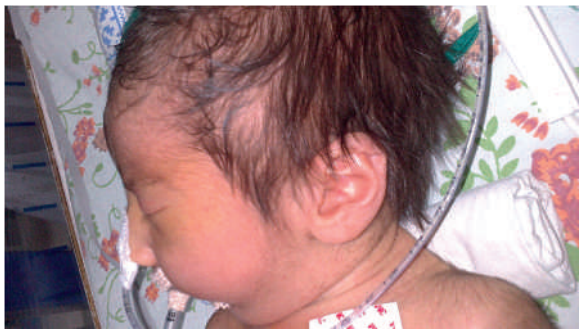

Fig 1: Facial Profile of Pierre Robin Sequence

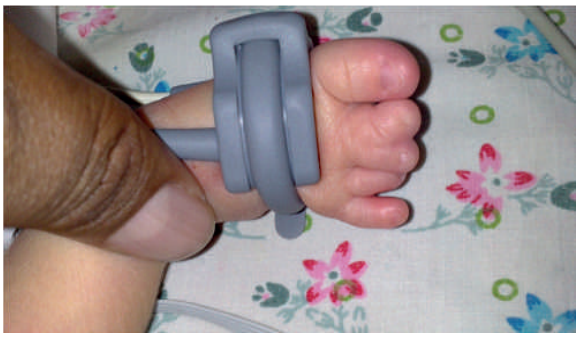

Fig 2: Syndactyly of right toe

The tongue is usually of normal size, but the floor of the mouth is shortened. Obstruction of the air passages may occur particularly on inspiration, and usually requires treatment to prevent suffocation. The infant should be maintained in a prone or partially prone position so that the tongue falls forward to relieve respiratory obstruction. When positioning alone fails, tongue base airway obstruction may be relieved by placement of a nasopharyngeal airway (NPA) without anesthesia. Some patients may require endotracheal intubation or rarely tracheostomy. Surgical procedures include tongue-lip adhesion (TLA), mandibular distraction osteogenesis (MDO), and tracheotomy[9]. Mandibular distraction procedures in a neonate can improve mandibular size, enhance respiration, and facilitate oral feedings[10]. Sufficient spontaneous mandibular growth may take place within a few months to relieve the potential airway obstruction. Often the growth of the mandible achieves the normal profile in 4 – 6 years. The feeding of infants with mandibular hypoplasia requires great care and patience but can usually be accomplished without resorting to gavage. Dental anomalies usually require individualized treatment[9]. The palatal cleft interferes with nursing and causes regurgitation of food through nose. Infection of the nasopharynx is frequent. Otitis media may result in 30- 40% of the afflicted, leading to hearing impairment or permanent deafness[1]. Bronchitis and pneumonia can complicate the local infections. In general, prognosis is good in majority of cases and death from PRS is thought to be the result of poorly controlled combined effects of obstructive apnea and failure to thrive.

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