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Case Report

Pierre Robin sequence in association with tracheo-oesophageal fistula: a heterogenous birth defect

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ABSTRACT

Pierre Robin Sequence (PRS) is not a genetic syndrome. It consists of multiple defects that occur as a result of a single structural anomaly. A full term male baby with micrognathia, retrognathia, glossoptosis and cleft palate was brought at second day of life for respiratory difficulty and unswallowed saliva. After clinical examination and abdomen radiograph, it was found that he had trachea-esophageal fistula. The fistula was ligated and esophageal anastomosis was done. Gavage feeding was commenced from fifth post-operative day with no problems. Karyotyping came out to be normal for him. On day 40 of life he arrived with failure to thrive and noisy breathing. Infants with Pierre Robin sequence develop airway obstruction soon after birth. Persistence of upper airway obstruction, is associated with serious consequences. This can be avoided by clinical evaluation and polysomnography.

Keywords: Micrognathia, Retrognathia, Cleft palate, Trachea-oesophageal fistula

INTRODUCTION

Pierre Robin sequence consists of multiple defects that occur due to single structural anomaly. It is described as a birth condition that involves the lower jaw being either small in size (micrognathia) or set back from the upper jaw (retrognathia). Due to which, the tongue tends to be displaced back towards the throat, where it can fall back and obstruct the airway (glossoptosis). Most infants have a cleft palate deformity. A French physician, Pierre Robin first reported small lower jaw, cleft palate and tongue displacement in 1923.

CASE REPORT

A two day old male baby weighing 2.5 kg, born at full term through normal vaginal delivery to a primigravida mother. He was admitted with complain of respiratory difficulty, feeding issues, unswallowed saliva. He had a cleft palate, micrognathia, retrognathia, glossoptosis and rocker bottom feet. Physical examination revealed dehydration and crepitations at right thorax. Oro-gastric tube passed through the mouth into the stomach to rule out esophageal atresia met with obstruction. A plain radiograph revealed that the catheter had turned around itself, suggesting the oesophageal atresia. A plain abdominal radiograph showed normal gas distribution, suggesting a distal trachea-esophageal fistula. Preoperative intubation was difficult due to the presence of micrognathia and glossoptosis. Under general anesthesia, the fistula was ligated and oesophageal anastomosis was performed. On fifth post-operative day, no anastomotic leakage was observed and gavage feeding was given for two weeks and maintained by a nasogastric tube were followed by a gastrostomy. Cleft palate repair was planned at the age of 9 months. Mandibular distraction was delayed because of gastro-oesophageal reflux, noisy breathing, respiratory problems. A venous

blood sample was obtained for genetic analysis. The cytogenetic results revealed a 46XY normal constitutional karyotype. The baby was followed up at the age of 40 days and had poor weight gain and his parents complained of intermittent noisy breathing. As he did not gain weight, he was advised and counselled for respiratory evaluation and polysomnogram, but parents refused because of financial constraints.

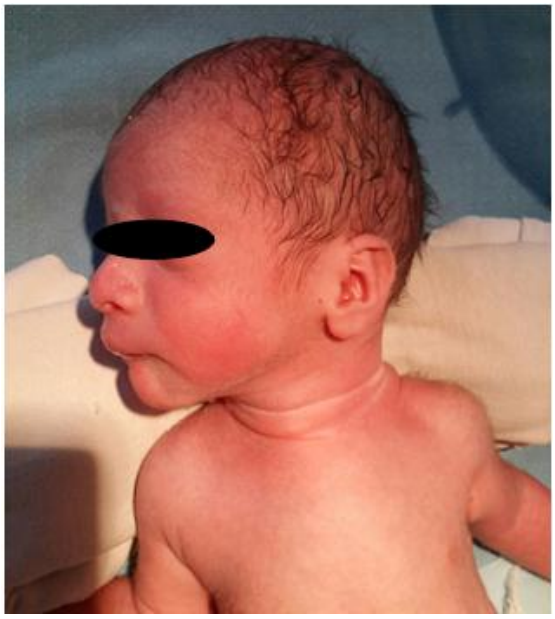

Figure 1: Neonate with retrognathia.

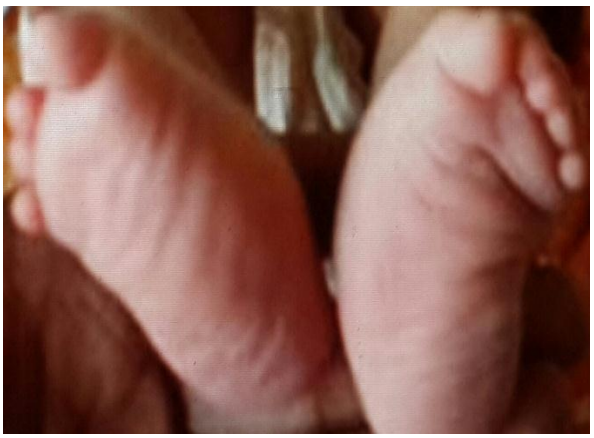

Figure 2: Neonate with rocker bottom foot.

DISCUSSION

PRS has a autosomal recessive type of inheritance.[1] Its prevalence is approximately 1 per 8500 live births. The male-to-female ratio is 1:1.[2] PRS may occur in isolation, but it can also occur in association with underlying disorder or syndrome like Stickler Syndrome, Velocardiofacial syndrome, Treacher Collins syndrome.[3] Non-syndromic PRS may be due to dysregulation of both SOX9 and KCNJ2. PRS may be caused by genetic anomalies at chromosomes 2, 11 or 17.[1]

PRS is thought to occur due to mechanical compression of the mandible, genetic growth disturbance, teratogen exposure and growth arrest due to an in utero insult. It occurs due to failure of the lower jaw to develop normally before birth. At about 7-10 weeks of pregnancy, the lower jaw grows rapidly, allowing the tongue to descend between the two halves of the palate. If the lower jaw does not grow properly, the tongue can prevent the palate from closing, resulting in a cleft palate. It is noted that when mandible is too small it fails to accommodate the descent of the tongue into the mouth; consequently, the tongue remains positioned between the sides of the developing palate thus preventing complete fusion. The small or displaced lower jaw also causes the tongue to be positioned at the back of the mouth, possibly causing breathing difficulty at birth. Due to this sequencing of events, PRS is classified as a deformation sequence.[4]

The child may present with respiratory difficulty soon after the birth due to mocrognathia, glossoptosis. They may have feeding issues if there is associated cleft palate. Obstructive sleep apnoeas may also occur. Most common otic anomaly is otitis media. Failure to gastroesophageal reflux and esophagitis is also seen. They may have abnormalities of the extremities such as syndactyly, hypoplastic digits and accessory metacarpals.[5]

The goals of treatment in infants with PRS are focussed on breathing and feeding, optimizing growth and nutrition despite the persistence of breathing difficulties. Parents should know how to position their infant so that the problem can be minimized. They should not place the infant in supine position. If the infant is severely affected, positioning may not play a significant role. In that case, they are given specially designed device to protect airway and to facilitate feeding. Some of them may require a surgical procedure for correction of the deformity.

Children with severe micrognathia are found to have significant respiratory obstruction at birth, requiring a nasopharyngeal airway or intubation. Children with minor degrees of PRS can learn to feed with nipple that has a large hole at the top, bottles such as Haberman feeder, Meade Johnson cleft palate nurser. In severe PRS, due to the risk of aspiration feeding is done with nasogastric tube.

Breathing problems can be solved by stomach positioning in prone position or by nasopharyngeal airway. Treatment is prioritised according to the severity of compromised airway followed by extent of feeding difficulties. Many surgical procedures have been described, but tracheostomy is most widely used. If there is glossopexy it should be relived before the development of dentition.[6]

The children can have late presentation of airway obstruction.[7] Failure to thrive in children with PRS is thought to be due to airway obstruction.[8] The association of PRS with oesophageal atresia need further clinical observations to clarify the underlying etiopathogenesis.

To conclude primary management of airway insufficiency in patients with PRS can be managed in a prone position mostly however some may require nasopharyngeal airway placement, prolonged intubation, surgical procedures and tracheostomy. Clinical evaluation alone is not sufficient to predict whether the children with PRS will develop airway obstruction or not, which suggest that there is a need of polysomnogram as a diagnostic tool for PRS. Multidisciplinary care that includes a neonatologist, a neonatal paramedical staff, oto-rhinolaryngologist team and the parents is the best approach in the complex care of neonates affected with PRS.

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