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Case Report

Pierre Robin syndrome: a case report

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ABSTRACT

Pierre Robin syndrome is characterized by micrognathia, glossoptosis and palatal malformation. We report a case of a 6 day neonate who presented with complaints of feeding and respiratory difficulty and was later diagnosed as case of Pierre Robin syndrome.

Keywords: Cleft palate, Micrognathia, Glossoptosis, Pierre Robin syndrome

INTRODUCTION

Pierre Robin Syndrome (PRS) is characterized by triad of micrognathia, glossoptosis and cleft palate. 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]

In 1923 French physician Pierre Robin introduced the term 'glossoptosis' in association with micrognathia. In 1934 he reported an association with cleft palate and this constellation of findings was later termed as syndrome. The Pierre-Robin Syndrome (PRS) is a rare malformating pathology and its estimated frequency is approximately 1/30000.[2] Some familial cases have also been reported, which may indicate that some cases have an inherited basis.

CASE REPORT

We report a case of 6 day old neonate gestational age 36 ± 2 weeks/(2.2 kg) LBW (Low Birth Weight) with AGA (appropriate for gestational age) born to 22 year old mother. The patient presented to Department of Paediatrics, S.S. Medical College, Rewa (M.P.) with the symptoms of feeding difficulty with respiratory distress.

On eliciting thorough detailed history it was revealed that mother had complaint of hyperemesis gravidarum in first trimester. On performing antenatal ultrasonography, oligohydramnios was detected with Amniotic Fluid Index (AFI) 6.

On careful examination of the patient we observed micrognathia, "U" shaped cleft palate and glossoptosis (Figure 1, Figure 2, Figure 3). Head circumference was 32 cm, length 44 cm, chest circumference 29 cm and new ballard score 32.

On respiratory system examination mild intercoastal and substernal recessions were seen. On CNS examination neonatal reflexes were sluggish. On cardiovascular examination no murmur was appreciated. Septic screening was normal. Two dimensional echocardiography was found to be normal. Opthalmological examination was found to be normal.

Patient gets relieved on feeding in prone or lateral position. No artificial airway was required in our case.

No family history was reported and he was first birth order with no siblings.

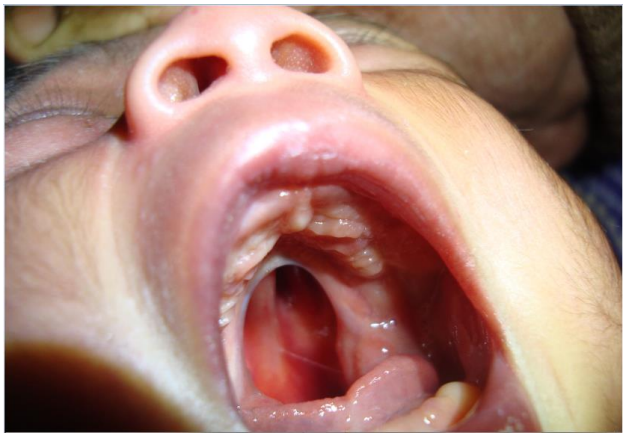

Figure 1: Showing U shaped cleft palate.

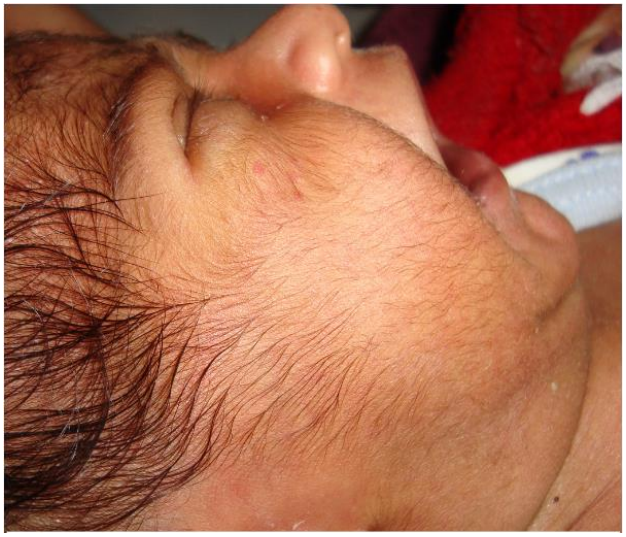

Figure 2: Showing micrognathia.

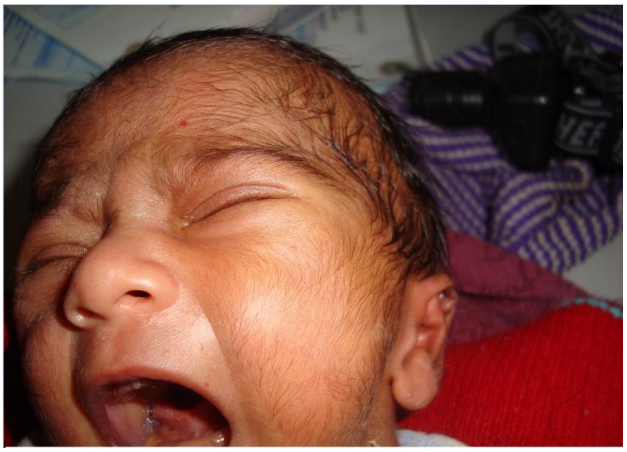

Figure 3: Showing glossoptosis.

DISCUSSION

Pierre Robin syndrome also termed as Pierre Robin sequence, or Robin anomalad, is characterized by several degrees of micrognathia severity, glossoptosis and palatal malformation. In this malformation, the tongue size is normal but buccolingual disproportion due to micrognathia, increasing the glossoptosis is frequently observed.

The word "sequence" suggests that one anomaly leads to subsequent anomalies, and micrognathia is considered to be the inciting anomaly in patients with PRS. Airway obstruction and feeding difficulties arise and the severity of the problems vary.

PRS may occur alone or in association with other syndromes such as stickler syndrome, velocardiofacial syndrome hence geneticist should be consulted to know the probability in future children. In about 30% of cases PRS may be an isolated occurrence, while, in the following 30%, it is related to other anomalies and in the last third of cases it is part of a more complex syndrome (most frequently Stickler Syndrome). This multiplicity of expressions is the result of a mixed genetic origin: in 40% of cases PRS is genetically isolated, otherwise it is a recessive or dominant autosomal condition.[3] In our case no association with other anomalies were observed.

Regardless of the cause, neonates and infants with Pierre Robin sequence may experience varying degrees of airway obstruction and feeding difficulties.[4,5] PRS can be life threatening during the neonatal period with the onset of airway obstruction, which can occur at any time right after birth. If left untreated, prolonged airway obstruction can lead to acute or chronic hypoxia, cyanosis, apnea episodes, aspiration, respiratory tract infection, feeding difficulties, malnutrition, and failure to thrive. Subsequent complications of chronic hypoxia are chronic carbon dioxide retention, elevated pulmonary vascular resistance, cor pulmonale, right heart failure, and cerebral hypoxia.[6-8]

Most of the patients with PRS gets relieved by conservative measures however patients with pronounced micrognathia, failure to thrive, prolonged use of nasopharyngeal airway or prolonged endotracheal intubation needs surgical correction. As in our case conservative measures were sufficient to manage the symptoms of neonate.

CONCLUSION

All the cases of PRS should be thoroughly investigated to diagnose association with other syndrome and to formulate the further line of management. Each and every case of PRS is unique and needs to be assessed individually. It is our responsibility as physicians to recognize this disorder timely, to provide close follow-up and appropriate therapy and counselling.

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