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Case series on Pierre Robin Syndrome

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Abstract

Background: Pierre Robin syndrome (PRS) is characterised by micrognathia (undersized jaw), glossoptosis (retracted tongue), and airway obstruction. Neonates with a complete type of cleft palate, frequently present with feeding problems and aspiration is a common compliction. Presented here are two cases with Pierre Robin Syndrome. *Case summary:* First case is a 3 month male with feeding difficulty and aspiration pneumonia had receding chin and cleft palate on examination. He was feed by nasogastric tube initially and later a obturator was fitted. Second case is a 6 month female with bronchpneumonia and had a high arched, U shaped cleft palate and a large tongue. *Conclusion:* Early diagnosis for proper feeding and growth of children with Pierre Robin Syndrome is important to prevent complications like failure to thrive and repeated aspiration pneumonia.

Keywords: Pierre Robin Syndrome; Micrognathia; Cleft Palate.

Introduction

The term 'glossoptosis' was first used by Pierre Robin, a French physician in 1923, in association with micrognathia. It was discovered later in 1934 that it is also association with cleft palate and thus termed as syndrome. Cohen renamed it almost 42 years later as anomalad which is 'a malformation with its subsequent-derived structural changes [1]. Currently, the term Pierre Robin sequence (PRS) is being used. Pierre Robin sequence (PRS) consist of micrognathia (undersized jaw), glossoptosis (retracted tongue), and upper airway obstruction, which has incidence of 1/8500 to 1/14,000 live births and is frequently associated with cleft palate [2,3]. Patients of Pierre Robin Syndrome are more prone to have upper respiratory airway obstruction or difficulty in feeding secondary to micrognathia i.e receding mandible, glossoptosis, or a posteriorly placed tongue which is in close proximity with the posterior pharyngeal wall [4].

Case report

Case 1

A 3 month male child was brought by his parents to our OPD with compliants of difficulty in feeding, cough and fever since 10 days. His birth weight was 2.3 Kg. He was born by vertex vaginal delivery with no significant antenatal maternal history. No significant family history. His past history was not significant. On head to toe examination, the neonate had a receded or undersized chin and a bird face appearance which is characteristic. Examination of oral cavity revealed a large cleft palate of complete type. Thus a tentative diagnosis of PRS was established. The diagnosis of PRS was made on the basis of findings of micrognathia and cleft palate of complete type and posteriorly placed tongue as backside of the tongue was visible by laryngoscopic examination. On examination of Respiartory system there were subcostal retractions and bilateral crept on auscultation of respiratory

system. The child was treated for pneumonia with a 7 day course of #3rd generation cephalosporins. To avoid the complications such as regurgitation or aspiration, as well as to facilitate feeding, it was decided to fabricate a palatal feeding obturator till the corrective surgery of cleft palate is undertaken.

Case 2

6 month child brought to OPD with complaints of fever and cough since 3 days. Parents gave a histroy of repeated upper respiratory tract infections in last 6 months. Also parents gave history of disturbed sleep pattern of child. She was fifth issue of non-consanguineous marriage born at term gestation by vertex vaginal delivery. Anthropometry and vitals were normal. On head to toe examination, she had high arched, U shaped cleft palate, micrognathia and a large tongue. She was also having CTEV of the right foot. No abnormality was found on systemic examination.

Fig. 1: Retrognathia and bird like facies

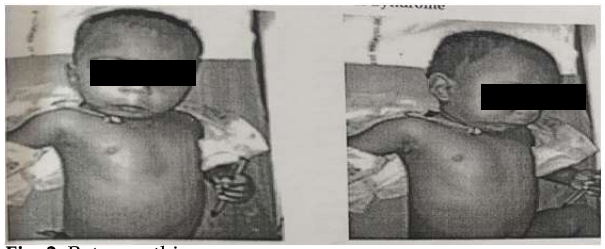

Fig. 2: Retrognathia

Discussion

Lannelongue and Menard in year 1891 discussed Pierre Robin syndrome in detail for first time by reporting 2 cases with micrognathia, cleft palate, and retro glossoptosis. The term 'glossoptosis' was first used by Pierre Robin, a French physician in 1923 in association with micrognathia. Later, in 1934, he found its association with cleft palate and this group of disorders was termed as syndrome. Later, in 1976, Cohen renamed this group of disorders as anomalad which is 'a malformation associated with its subsequent-derived structural changes [5].

The exact pathogenesis of the physical abnormalities remain uncertain but according to one theory, at some point of time during the development of the bones of the fetus in gestational period, the tip of the jaw (mandible) becomes 'stuck' in the point where each of the collar bones (clavicle) meet (the sternum) effectively, thus hampering further growth of the mandible. Approximately around 12 to 14 weeks of gestational period, it is observed that the fetus starts to move, and the subsequent movement of the head results in the jaw to "pop out' of the collar bones or clavicles [6]. The mandible of the developing fetus then grows as it would have normally been, but with the result that, at birth, the mandible of the baby is much smaller (micrognathia) than expected with normal development. But mandible continues to develop at a normal rate of development until the child reaches maturity.

Also, association of the syndrome with gene loci 2q24.1-33.3, 4q32-qter, 11q21-23.1, and 17q21-24.3 has been found. In recent studies, it has been found that genetic dysregulation of SOX9 gene causes prevention of the SOX9 protein from properly controlling the development of facial structures, which leads to isolated PRS. Similarly, KCNJ2 gene has also found to have a role to play. Overlap with certain other genetic syndromes like Patau syndrome has also been found [7].

This heterogeneous birth defect has a prevalance of 1:8500 live births with a male:female ratio of 1:1. Micrognathia is reported inmajority of cases (91.7%). It is characterised by retraction of inferior dental arch 10-12 mm below the superior dental arch. The mandible has a small body, Obtuse genial angle and a posteriorly located condyle.

Glossoptosis is observed to present in 79-85% of reported cases. Macroglossia I.e large tongue and ankyloglossia are less frequently observed. Obstructive sleep apnoea is possibility in such cases. The most common Otic anomaly is recurrent otitis media. Conductive hearing loss is common while ear canal atresia occurs in very few patients. Nasal anomalies are less frequently seen and

consist mostly of nasal root anomalies. Difficulty in speech occur commonly in patients with Pierre Robin syndrome due to the facial anomalies [8,9].

The cause of death in these patients is mostly obstructive apnea due to airway blockage, aspiration peumonia and failure to thrive. The palatal cleft causes difficulty in feeding and causes regurgitation of food through nose. Otitis media may result in approximately 50% of the patients and it can lead to hearing permanent deafness.

Bronchitis and pneumonia are frequent complications noted. The construction of the palatal obturator or a feeding plate is used to prevent the regurgitation of food. Newer treatment modalities consist of the use of specially designed nipples with enlarged openings, and use of orogastric and naso gastric tubes. Feeding obturator is a prosthetic aid that is designed to obturate the cleft and makes separation on the oral and nasal cavities possible [10].

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