

CASE REPORT

PIERRE ROBIN SEQUENCE: A RARE PRESENTATION OF ABSENT FEMUR AND INGUINAL HERNIA

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Pierre Robin Sequence is a well-recognized case of rarity which presents with a triad of glossoptosis, cleft palate. We report a rare case of Pierre robin Sequence in an Asian child, with the absence of left femur and right sided inguinal hernia.

Keywords: Pierre Robin, cleft palate, Absent Femur, inguinal hernia

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INTRODUCTION

Pierre Robin sequence (PRS) is a congenital anomaly that presents with a border group of anomalies which manifests as the isolated defect, which may present as microganathia, glossoptosis & cleft palate. In 1934, Pierre Robin, a French physician proposed this rare condition in which he first time proposed the term microganathia, where as in 1976, he added the term cleft palate and proposed it as Pierre Robin Sequence.¹ The major defect is the arrested growth of the mandible which leads to the appearance as “bird shape appearance of face “which ultimately leads to absence of descent of tongue between the palatal shelves. Most of the parents complain as the hindrance faced during feeding at the time of the birth and child turns. The prevalence of PRS was found from 1/8000 to 1/14,000 births.² A case report has been reported in the recent past where few other anomalies have also been reported along with the Triad of PRS such as absence of proximal femur, radio styloid synostosis and micro gastria which are rare presentation.³

CASE REPORT

Parents of 9 months old child reported to Paediatric department of Abassi Shaheed Hospital with the chief complain of high grade fever, difficulty in breathing.

On GPE we found the child restless & irritable with the characteristic appearance of receded chin, bird shape faces (Figure-1a, 1b, 1c) & absence of the left side proximal femur (Figure-2a), patient also has right sided inguinal hernia which was non- reducible, soft on examination (Figure-2b)

Intramural examination of the child revealed large cleft normal appearing gum pad & small lower jaw, based on these findings, a diagnosis of cleft palate was made by the senior paediatricians & plastic surgeons. Figure-3a.

Neck examination revealed glossoptosis creating difficulty in breathing. Chest examination of the child revealed centrally compressed sternum, intercostal recessions, on auscultation bilateral wheezes were present. Remaining examination was non-significant.

Lab report revealed high TLC of 16,000, bold culture reported presence of s .pneumonia, we managed the child by admitting to ICU , patient was given ceftriaxone 500mg I/V od for 5 days along with the conservative management for fever, child responded well to antibiotics & then shifted to ward where Nasogastric tube was passed .After complete recovery, patient was shifted to the department of plastic surgery for evaluation and plan for surgery of cleft palate, for this purpose child was intimated by the department of Ear Nose and Throat.



Figure-1a



Figure-1b



Figure-2a

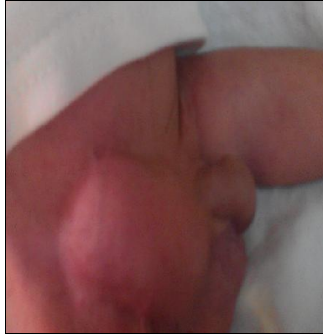


Figure-2b



Figure-3a



Figure-3b

DISCUSSION

Pierre Robin syndrome is characterized by the of the triad of micrognathia, glossoptosis & labiopalatine clefting, along with few rare associations, we report a case of PRS from Pakistan where prevalence of Pierre Robin sequence is extremely low, along with the rare association of absent left femur bone as shown in x-ray. Up to best of our article search revealed only one case report of PRS knowledge in a child with oligodactyly.⁴

Generally deaths from PRS (Pierre Robin Sequence) result from combined factors of Obstructive sleep apnoea or failure to thrive. palatal cleft interferes with the feeding & results in the regurgitation of fluid from the nose leading to the infection of nose, few patients also suffer with the otitis media leading to the hearing impairment⁵ with the overwhelming infections which lead to the complications of pneumonia and bronchitis, development of the palatal obturator or feeding plate or a widely designed feeding nipple or use of or gastric or nasogastric tubes which are used mostly in weaker infant.⁶

Evaluation of air way is must, therefore sleep and feeding monitoring of spontaneous oxygen desaturation and during phonation is done, furthermore endoscopy & bronchoscope are invaluable to be done to determine site of obstruction.⁷

Tongue lip adhesion is the procedure which correct glossoptosis by stretching forward of tongue base & suturing it to lower lip .The procedure serves to correct the problem of glossoptosis by pulling the base of the tongue forward and suturing it to the lower lip, once healed the patient can develop suitable growth of the air ways decreasing the obstruction in the airway.⁸

Distraction ontogenesis (DO) of the mandible is another treatment method, first described in 1989 by McCarthy, done in patients with PRS for relieving airway obstruction, improving facial, This procedure

lengthens the jaw in a forward direction, and indirectly pulls the tongue base anteriorly, the tongue is also pulled anteriorly through its muscular attachments with time as the mandible moves forward.⁹

Feeding obturator is a device that used to obturate in between cleft and restore place between the oral & nasal cavities.⁶

CONCLUSION

Pierre robin sequence is a rare genetical disease worldwide, and is reported more rarely in countries of South East Asia; hence we report a case report of Pierre Robin Sequence from Pakistan along with association of absence of left femoral bone.

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