Pierre Robin Syndrome

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Abstract :

Pierre Robin syndrome a well-recognized congenital condition characterized by micrognathia, glossoptosis and palatal malformation . Pierre Robin syndrome is also called pierre Sequence because it is caused by sequence of events that happen in the womb , which affects the way the baby develops. We report a case of a one day neonate who presented with complaints of feeding and respiratory difficulty and was later diagnosed as case of Pierre Robin syndrome. The primary defect lies in the arrested development of mandible leading to a characteristic bird face appearance.

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 syndrome is also called pierre Sequence because it is caused by sequence of events that happen in the womb , which affects the way the baby develops. Pierre Robin Syndrome 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. Some familial cases have also been reported, which may indicate that some cases have an inherited basis.[2]

Pierre Robin Syndrome may occur alone or in association with other syndromes such as stickler syndrome, velo-cardio-facial syndrome. In about 30% of cases, Pierre Robin Syndrome 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).[3]

The restricted growth of the babys jaw is the first event in the sequence, as consequence, space in the babys mouth becomes very limited, so that babys tongue prevents the palate from closing and can partially block the airway.[3]

Case Report:

Fig. 1
Micrognathia

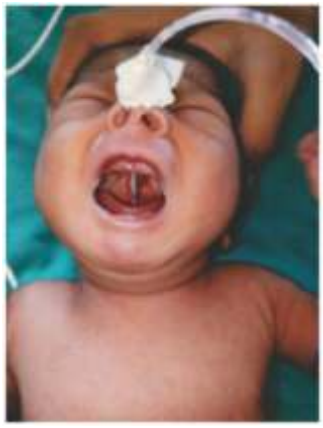

Fig. 2
Cleft Palate & Microglossia

We report a case of one day old neonate , gestational age 36 ± 5 weeks/ (2.2 kg, Low Birth Weight) with AGA, born to 22 year old mother with history of oligo-hydramnios and with breech presentation.

The patient presented to our Department of Paediatrics 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 ultra-sonography, oligo-hydramnios was detected with Amniotic Fluid Index (AFI- 4).

On careful examination of the baby, baby has micrognathia, "v" shaped cleft palate and glossoptosis (Figure 1, Figure 2). Head circumference was 32.5 cm, length 46 cm, chest circumference 28 cm and Modified ballard score 36.

On respiratory system examination mild intercoastal and substernal recessions were seen. On CNS examination neonatal reflexes were normal. On cardiovascular examination , murmur was appreciated. Septic screening was normal. Two dimensional echocardiography was found, Interatrial septal aneurysm and Persistent foramen ovale (PFO) of size 2mm. Opthalmological examination was found to be normal.

No artificial airway was required in our case. No family history was reported and he was first birth order with no siblings. In our case no association with other anomalies were observed.

Discussion : Lannelongue and Menard first described Pierre Robin syndrome in 1891 in a report on 2 patients with micrognathia, Cleft palate, and retroglossoptosis. 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. In 1976, Cohen renamed it as anomalad which he defined as 'a malformation together with its subsequent-derived structural changes.[4]

It is not known how this abnormality occurs in infants, but one theory is that, at some time during the stage of the formation of the bones of the fetus, the tip of the jaw (mandible) becomes 'stuck' in the point where each of the collar bones (clavicle) meet (the sternum), effectively preventing the jaw bones from growing. It is thought that, at about 12 to 14 weeks gestation, when the fetus begins to move, the movement of the head causes the jaw to "pop out' of the collar bones. From this time on, the jaw of the fetus grows as it would normally, with the result that, when born, the jaw of the baby is much smaller (micrognathia) than it would have been with normal development, although it does continue to grow at a normal rate until the child reaches maturity.[5]

In general, death from PRS is thought to be the result of poorly controlled combined effects of obstructive apnea and failure to thrive. 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.

Bronchitis and pneumonia can complicate the local infections. Hence, the construction of the palatal obturator or a feeding plate is very important until the surgical correction of the defect is carried out. Other treatment modalities include the use of specially designed nipples with enlarged openings, and use of orogastric and naso gastric tubes.[6]

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