Pierre Robin sequence in association with tracheoesophageal fistula and esophageal atresia

Yusuf Atakan Baltrak[1], Onursal Varlikli[2]

From Specialist Pediatric Surgery, Departments of Pediatric Surgery, [1]Kocaeli Derince Education and Research Hospital, Derince, Kocaeli, [2]Sakarya Education and Research Hospital, Sakarya, Turkey

Correspondence to: Yusuf Atakan Baltrak, Departments of Pediatric Surgery, Kocaeli Derince Education and Research Hospital, Derince, Kocaeli, Turkey. E-mail: dratakanbaltrak@gmail.com

Received - 19 February 2019 Initial Review - 15 March 2019 Accepted - 26 march 2019

ABSTRACT

The first symptom of esophageal atresia and tracheoesophageal fistula is the inability to eat and respiratory distress after feeding. The coexistence of Pierre Robin Sequence (PRS) with esophageal atresia and tracheoesophageal fistula is a rare clinical condition. In infants with PRS, evaluation of respiratory and nutritional problems is important. The coexistence of esophageal atresia and tracheoesophageal fistula leads to increased nutritional and respiratory problems. Problems that may occur in the airways may occur late. In this case report, we aimed to present a case of neonatal infant with rare esophageal atresia, tracheoesophageal fistula, and PRS.

Key words: *Esophageal atresia, Pierre Robin Sequence, Newborn*

Pierre Robin's sequence (PRS), described for the 1st time by a French doctor Pierre Robin in 1923, manifests itself with a single structural anomaly and more than one clinical symptom. In the postpartum examination, it is observed that the lower jaw is small (micrognathia) and is positioned relatively behind the upper jaw (retrognathia); the tongue is placed backward toward the throat obstructing airway (glossoptosis) and there is also cleft palate deformity. Esophageal atresia occurring together with PRS is a rare condition. In this case report, we aimed to present a rare case of esophageal atresia, tracheoesophageal fistula, and PRS in neonatal infants [1].

CASE REPORT

Our case was delivered out of a normal spontaneous vaginal delivery with a body weight of 3435 g, a head circumference of 34 cm, and a height of 50 cm, from the 2nd pregnancy of a 28-year-old mother as the second live birth. In the patient's first physical examination performed in the delivery room, the patient was thought to have Pierre Robin Sequence after micrognathia, glossoptosis and cleft palate were observed (Fig. 1a-c). The patient turned blue after the first feeding and had plenty of mucus in the mouth. The patient was considered to have esophageal atresia, and orogastric tube was tried to be descended into the stomach. Esophageal atresia was suspected because the orogastric tube could not be advanced >12 cm. When X-ray was taken with contrast material, atresic proximal esophageal pouch was detected (Fig. 2a). The patient was diagnosed with esophageal atresia and tracheoesophageal fistula due to the gas in the stomach and intestines. Abdominal USG for additional anomaly screening was reported as normal. Cardiac echocardiography was reported as "pathological demand avoidance and seguinum autism spectrum disorder may be operated under infective endocarditis prophylaxis." The patient with cleft palate and micrognathia was evaluated for difficult intubation by anesthesia. The patient's Pierre Robin gene was sent for analysis. The result was positive for Pierre Robin Sequence.

On the postnatal 1st day, as a result of the exploration on patient who underwent surgery for esophageal atresia and tracheoesophageal fistula, tracheoesophageal fistula was seen on tracheal bifurcation line (Fig. 2b). The patient underwent tracheoesophageal fistula repair and primary repair of esophageal atresia and was discharged on the 8th post-operative day due to cleft palate with feeding recommendations. In the post-operative follow-up, 1 month after operation, no problem has been observed until now. In this case report, written and oral consent was obtained from the patient's family.

DISCUSSION

PRS has an autosomal recessive inheritance type. Its prevalence is approximately 1 in 8500 live births. Male-female ratio is 1:1 [1,2]. PRS may be caused by a genetic abnormality in chromosome 2, 11, or 17, or it may be caused by an isolated or underlying cause. It may be associated with a disorder or syndrome such as syndromic PRS Stickler syndrome, velocardiofacial syndrome, and Treacher Collins syndrome,

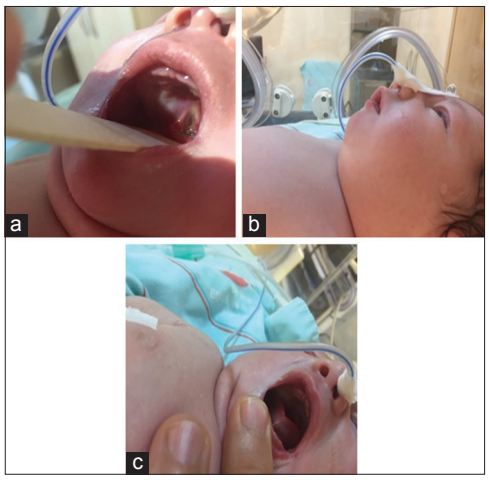

Figure 1: (a) Cleft palate, (b) Micrognathia, (c) Glossoptosis

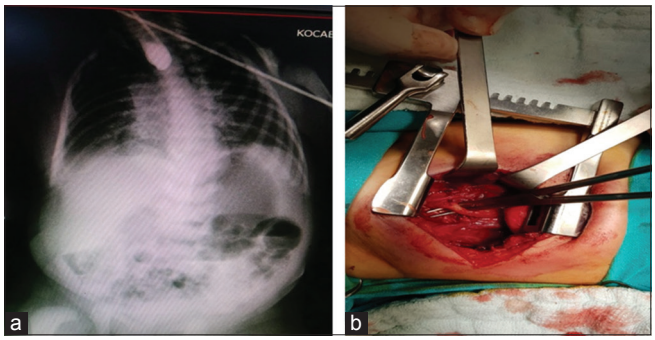

Figure 2: (a and b) Atretic proximal esophageal pouch and tracheoesophageal fistula

while non-syndromic PRS may be caused by a disorder in SOX9 and KCNJ2 genes [1,3].

PRS is thought to be caused by compression of mandible during intrauterine period, teratogenic exposure, and genetic growth disorder. Micrognathia is thought to occur before birth. The lower jaw grows rapidly between 7th and 10th weeks of intrauterine life. If the jaw does not grow properly, the closure defect, which results in cleft palate, occurs in the palate. When the mandible is too small, the downward descent of the tongue is restricted. Due to the small and backward placed lower jaw, the tongue that is positioned backward in the mouth may cause respiratory distress [4]. The child may suffer respiratory distress due to micrognathia and glossoptosis. If there is a cleft palate associated with them, the patient may suffer with feeding problems. It may be accompanied by obstructive sleep apnea. The most common ear anomaly is otitis media. Gastroesophageal reflux and esophageal reflux can also be seen. Extremity anomalies, syndactyly, and hypoplastic fingers may accompany this [5].

Treatment in PRS infants is focused on overcoming respiratory difficulties and feeding problems. Parents can minimize these problems by keeping babies in a proper position. The baby should not be placed in the supine position. In case of severe feeding problem and respiratory distress, a specifically dedicated device is used to facilitate feeding and breathing. In some cases, surgery may be necessary to correct the deformity [6]. It is important to evaluate respiratory and feeding problems in babies with PRS and esophageal atresia. The occurrence of respiratory problems (if any) may arise later [7]. Development retardation in PRS babies is thought to be due to respiratory problems [8].

CONCLUSION

The solution of respiratory problems that may arise in patients with PRS is not difficult and can be easily treated with few measures. It includes lying in prone position, using nasopharyngeal respiratory device and long-term intubation and tracheostomy tube opening can also be considered.

REFERENCES

1. Robin P. Glossoptosis due to atresia and hypotrophy of the mandible. Am J Dis Child 1934;48:541-7.
2. Gruen PM, Carranza A, Karmody CS, Bachor E. Anomalies of the ear in the Pierre Robin triad. Ann Otol Rhinol Laryngol 2005;114:605-13.
3. Elzen AP, Semmekrot BA, Bongers EM, Huygen PL, Marres HA. Diagnosis and treatment of the Pierre Robin sequence: Results of a retrospective clinical study and review of the literature. Eur J Pediatr 2001;160:47-53.
4. Thakur GV, Kandakure VT, Thote A, Ayesha K. Pierrie Robin syndrome-case review. Int J Sci Res Public 2013;3:1-4.
5. Farnsworth PB, Pacik PT. Glossoptotic hypoxia and micrognathia. The Pierre Robin syndrome reviewed. Clin Pediatr 1971;10:600-6.
6. Bath AP, Bull PD. Management of upper airway obstruction in Pierre Robin sequence. J Laryngol Otol 1997;111:1155-7.
7. Ogborn MR, Pemberton PJ. Late development of airway obstruction in the Robin Anomolad (Pierre robin syndrome) in the new-born. Aust Paediatr J 1985;21:199-200.
8. Dennison WM. The Pierre Robin syndrome. Pediatrics 1965;36:336-41.

Funding: None; Conflict of Interest: None Stated.

How to cite this article: Baltrak YA, Varlikli O. Pierre Robin sequence in association with tracheoesophageal fistula and esophageal atresia. Indian J Child Health. 2019; 6(4):196-197.

Doi: 10.32677/IJCH.2019.v06.i04.013