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Proper Interventions in a Newborn with Cerebro-Costo-Mandibular Syndrome

Serebro-Kosto-Mandibular Sendromlu Bir Yenidoğanda Uygun Girişimler

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ABSTRACT

Cerebro-costo-mandibular syndrome is characterized by severe micrognathia with glossoptosis, short to cleft soft palate, bell-shaped small thorax with gaps between the posterior ossified rib and anterior cartilaginous rib and postnatal growth and mental deficiency. We present the clinical course and the early aggressive treatment of a newborn diagnosed as cerebro-costo-mandibular syndrome. The management of the patients with this syndrome is important over the morbidity and mortality. Most babies died due to respiratory distress. Although brain anomalies are uncommon in cerebro-costo-mandibular syndrome, hypoxia may cause neurodevelopmental disorders. In this case report, we emphasize the importance of proper ventilation and feeding in patients with cerebro-costo-mandibular syndrome to maintain normal development. Multidisciplinary approach is essential in monitoring of these patients.

Keywords: Cleft palate, glossoptosis, laryngeal mask airway, palatal plate, respiratory insufficiency

ÖZ

Serebro-kosto-mandibular sendrom, glossopitosisin eşlik ettiği ağır mikrognati, kısa-yarık yumuşak damak, kotların arkadaki kemik kısmı ile ön kıkırdak kısımları arasında boşlukla karakterize küçük göğüs yapısı ve postnatal gelişme geriliği ve mental gerilik ile karakterizedir. Bu yazıda serebro-kosto-mandibular sendrom tanısı alan bir yenidoğanda klinik seyrin ve erken tedavinin önemini vurgulamak istedik. Bu hastalarda tedavi yönetimi morbidite ve mortalite üzerine çok etkilidir. Çoğu bebek solunum yetmezliği nedeni ile kaybedilmektedir. Serebro-kosto-mandibular sendromlu hastalarda beyin anomalileri sık olmamasına karşın nörogelişimsel sorunların oluşumunda hipoksi önemli rol alır. Bu vakada etkin ventilasyon ve uygun beslenmenin normal gelişimi sağlamada etkili olduğunu vurgulamak istedik. Bu hastaların izleminde multidisipliner yaklaşım esastır.

Anahtar Kelimeler: Yarık damak, glossopitozis, larengeal maske, palatal plak, solunum yetmezliği

Introduction

Cerebro-costo-mandibular syndrome (CCMS) is characterized by severe micrognathia with glossoptosis, short and cleft soft palate, bell-shaped small thorax with gaps between the posterior ossified rib and anterior cartilaginous rib and postnatal growth and mental deficiency (1, 2). It was initially reported in 1966 and to date, 60 cases have been reported (1).

The urgent problem of the disease is glossoptosis, which may cause respiratory insufficiency and hypoxia, resulting in death or neurodevelopmental disorders (3, 4). Feeding problems and its outcome failure to thrive is an additional problem in these babies. Cleft palate may cause aspiration, nasogastric tube complications, and inadequate feeding (5).

We here describe a case of CCMS with severe respiratory distress and discuss the proper interventions to improve the quality of life in these patients.

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Case Report

A male baby was born by spontaneous vaginal delivery at 39 weeks of gestation to a 20-year-old gravida 1, para 1 mother. The mother had oligohydramnios. The parents were first cousins. The birth weight was 3330 gram and length was 52 cm. Shortly after birth, he presented with signs of respiratory distress and central cyanosis. He was transferred to the neonatal intensive care unit.

On physical examination, he was found to have micrognathia with glossoptosis, high and large cleft palate. Chest radiography revealed gaps between the posterior ossified rib and anterior cartilaginous rib (Figure 1). The echocardiography showed patent foramen ovale, whereas the ultrasonography of the brain and the abdomen were normal. The analysis of the karyotype was normal.

Respiratory distress was initially managed with nasal oxygen supplement and prone positioning. The palatal plate was performed by the orthodontic department to manage feeding on the first day of life. On the 5th day of life, acute respiratory insufficiency was observed and the baby could not intubated. Laryngeal mask airway (LMA) was used and tracheostomy was planned. The baby was ventilated for 2 days by LMA. On the 7th day of life, tracheostomy was performed. After tracheostomy was performed, the baby was followed up at room air and the mechanical ventilation was ceased. He was transferred to plastic surgery to perform distraction osteogenesis of the mandible at 29th day of life.

The tracheostomy was closed when he was 2 years old. Now, the baby is 3 years old and there is no neurodevelopmental disorder.

Written informed consent was obtained from the patient's parents.

Discussion

The clinical picture of the CCMS in the newborn period depends on the Robin sequence (1, 6). Overall, 32% of infants died during the neonatal period because of respiratory distress (1). Prone positioning with nasopharyngeal tubes, LMA, tongue-lip adhesion operation, and tracheostomy were the emergent respiratory procedures (5, 7, 8).

Prone positioning is a noninvasive and an easy method to maintain an unobstructed airway. Nevertheless, in babies with big tongues like our patient, it is not safe enough at home. It does not maintain a long-term safe airway (8).

Laryngeal mask airway is preferred in neonatal resuscitation in babies with Pierre Robin syndrome. Intubation is difficult because of the anotomical structure of these anomalies. On the other hand, LMA cannot be used for longer durations because of the decreased perfusion of the pharyngeal mucosa and the risk of pharyngolaryngeal morbidity (5).

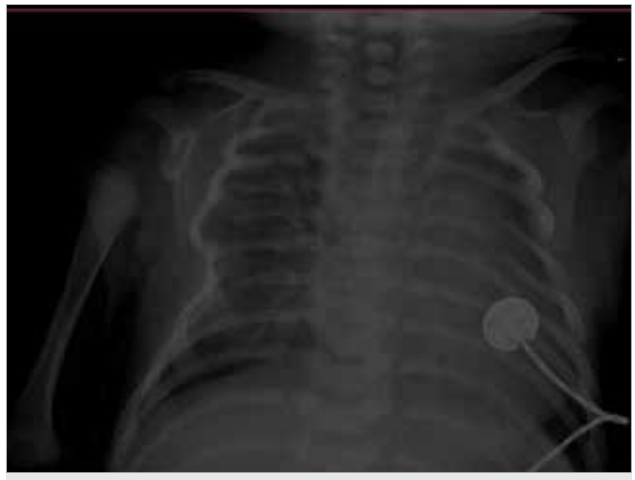

Figure 1. Chest radiograph reveals the gaps between ossified and cartilanegeus ribs

Some clinicians preferred tracheostomy (8, 9). Tracheostomy can ensure that the airway stays open until reconstructive surgery is performed. We also provide airway patency with tracheostomy without any complications.

Distraction osteogenesis of the mandible is the another treatment choice but it cannot be done during early life (5, 7). Wilcox and Tatum reported a patient with subglottic stenosis who required tracheostomy after osteogenic distraction (7).

Feeding and speech difficulties were seen in survivors. To provide optimum feeding, prosthetic palate was the beneficial approach until reconstructive surgery was performed (8). In a retrospective study, using the plates in early neonatal period seemed to decrease the requirement of the surgery, duration of hospitalization, and morbidity of palatal reconstruction (8). Nasogastric tube feeding and the gastrostomy were the other choices of feeding in these patients; however, feeding tube problems were also reported in these patients (7-9). Palatal reconstructions were performed at 18 months of life. However, palatal plate can be placed within the the initial days after birth. Also, these plates managed the respiratory distress and shortened the length of stay at the neonatal intensive care unit (8).

The management of the patients with CCMS is important over the morbidity and mortality. Most babies died because of respiratory distress. Although brain anomalies are uncommon in CCMS, hypoxia may cause neurodevelopmental disorders (9, 10).

Conclusion

In this case report, we emphasize the importance of the proper ventilation and feeding in CCMS patients to maintain the normal development. A multidisciplinary approach is essential in the monitoring of these patients.

Informed Consent: Written informed consent was obtained from patients' parents who participated in this case.

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