Clinical and Experimental Health Sciences

Case Report / *Olgu Sunumu*

Swallowing Therapy for a Case of Congenital Absence of the Epiglottis

Konjenital Epiglottis Yokluğu Olan Vakada Yutma Terapisi

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Abstract

The epiglottis plays an important role during swallowing to prevent food penetrating into the airway by closing down the laryngeal vestibule during laryngeal elevation. In case of the absence of the epiglottis, airway closure might fail. The absence of the epiglottis is rarely seen in patients with Pierre Robin Sequence (PRS). We report here the swallowing problems of a PRS case with absence of the epiglottis and the results of our interventions. A male infant who was born by cesarean section with a weight of 3 kilograms was diagnosed with PRS. When he was 11 months old, he was referred to Hacettepe University, Department of Physiotherapy and Rehabilitation, Swallowing Disorders Units due to a history of aspiration pneumonia. First, a fiberoptic endoscopic swallowing evaluation (FEES) was performed and the absence of the epiglottis was recognized. Because of the uncomfortable feeling during FEES, videofluoroscopic swallowing evaluation (VFSE) was also performed for a more detailed swallowing evaluation. Aspiration was seen. A nasogastric tube was inserted after VFSE, and a swallowing therapy program was followed for 2 months. Thermal tactile stimulation to trigger swallowing reflex, laryngeal mobilization to support hyolaryngeal elevation, and neuromuscular electrical stimulation were used. VFSE was repeated after rehabilitation. Aspiration of liquid consistency continued, but he tolerated pudding consistency. At that time, liquid-restricted oral intake was started. Early diagnosis and intervention is very important in patients with absence of the epiglottis.

Keywords: Pierre Robin Syndrome, epiglottis, deglutition, deglutition disorders

Öz

Epiglottis laringeal elevasyon sırasında vestibülü kapatarak, yutma esnasında hava yoluna besin kaçmasını engellemede önemli bir rol oynamaktadır. Epiglottisin olmadığı durumda, hava yolu kapanışı sekteye uğrayabilir. Pierre Robin Sendrom'lu (PRS) hastalarda epiglottis yokluğu nadir görülmektedir. Burada epiglottis'i olmayan bir PRS vakasını ve tedavi uygulamalarının sonucunu sunulacaktır. PRS teşhisi konmuş erkek infant 3 kg doğum ağırlığına ve sezaryen doğum hikayesine sahipti. Vaka 11 aylıkken aspirasyon pnömonisi hikayesi sebebiyle Hacetttepe Üniversitesi Fizyoterapi ve Rehabilitasyon Bölümü, Yutma Bozuklukları Ünitesine yönlendirilmiştir. İlk önce fiberoptik endoskopik yutma değerlendirilmesi (FEYD) yapılmış ve epiglottis yokluğu farkedilmiştir. FEYD sırasındaki rahatsızlık hissi nedeniyle, daha detaylı bir yutma değerlendirilmesi için videofluoroskopik yutma değerlendirilmesi (VFYD) gerçekleştirilmiş ve aspirasyon görülmüştür. VFYD'den sonra hastaya nazogastrik tüp takılmış ve 2 aylık bir yutma rehabilitasyon programına başlanmıştır. Program dahilinde yutma refleksini tetiklemek amacıyla termal taktil stimulasyon, hyolaringeal elevasyonu artırmak için laringeal mobilizasyon ve nöromuskuler elektrik stimulasyonu uygulanmıştır. Rehabilitasyon sonrası VFYD tekrarlanmıştır. Sıvı kıvamlarda aspirasyonun devem ettiği ancak puding kıvamının tolere edildiği görüldü. Değerlendirme sonrasında sıvı kısıtlı oral alıma başlandı. Epiglottisi olmayan hastalarda, erken teşhis ve tedavi yaklaşımları çok önemlidir.

Anahtar kelimeler: Pierre Robin Sendromu, epiglottis, yutma, yutma bozuklukları

INTRODUCTION

The congenital absence of the epiglottis is a rare condition. One of the most common causes of congenital absence of the epiglottis is Pierre Robin Sequence (PRS). PRS is a congenital condition characterized by micrognathia, glossoptosis, cleft palate, and laryngeal anomalies (1), and it causes feeding problems and respiratory complications. Although PRS is a common disease, the absence of the epiglottis, which is a laryngeal anomaly, is still rarely seen with PRS (2).

The epiglottis plays an important role in protecting the airway against food penetration by closing the laryngeal entrance during swallowing (3). Thus, the absence of the epiglottis increases the risk of aspiration.

Studies on the absence of the epiglottis are limited. This problem is usually diagnosed in infancy and early childhood. The first case with aplasia of the epiglottis who also had complete median cleft palate was reported in 1983 in *SA Medical Journal* (4). A 3-year-old boy and a 14-year-old girl with absence of the epiglottis were also reported in 1998 in the US (5, 6). Cases with hypoplastic epiglottis were also seen in previous reports. A 42-year-old man and a male infant with hypoplastic epiglottis have also been published as case reports (7, 8). Existing case reports defined the medical status, anomalies, their evaluation, and management of the patients. Swallowing problems were emphasized, but there is no study in the literature showing the swallowing rehabilitation outcomes in patients with absence of the epi-

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Figure 1. The case with Pierre Robin Sequence (PRS) demonstrating micrognathia

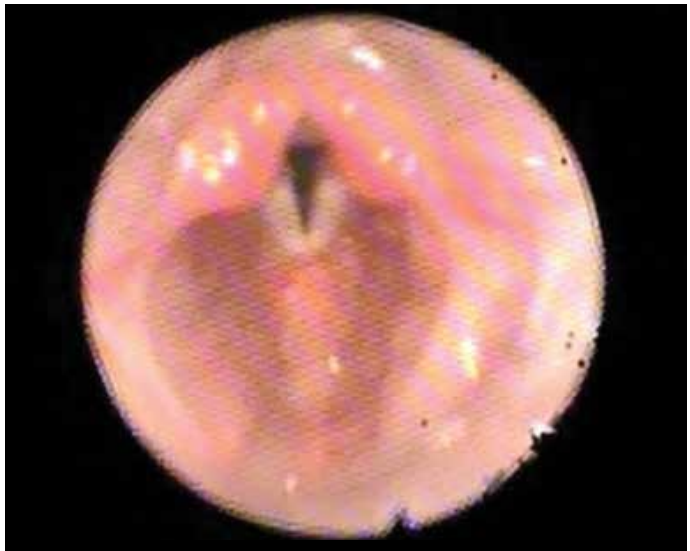

Figure 2. The absence of the epiglottis during Fiberoptic Endoscopic Swallowing Evaluation (FEES)

Table 1. The results of videofluoroscopic swallowing evaluation (VFSE)

	Before therapy (11 months old)		After therapy (13 months old)	
	Liquid	**Pudding**	**Liquid**	**Pudding**
Penetration Aspiration Score	8	8	5	1
Silent aspiration	+	+	–	–
Nasal reflux	+	+	–	–

Table 2. Body weight, height, and frequency of lung infections in the infant

	Before therapy (11 months old)		After therapy (13 months old)	
	Liquid	**Pudding**	**Liquid**	**Pudding**
Body weight (kg)	6.5	9.5	5	1
Height (cm)	65	72	–	–
Frequency of lung infection	11	0	–	–

glottis. We report here the swallowing problems of a PRS case with absence of the epiglottis, our intervention, and the results of the intervention. Case studies in the literature that exist are only case definitions, whereas in our case the rehabilitation results are discussed.

CASE PRESENTATION

The male infant was born by cesarean section with a weight of 3000 g. His parents were consanguineous, and his brother was diagnosed with autism. There were no congenital anomalies in the parents, and there were no maternal complications during pregnancy. Micrognathia and cleft palate were recognized at birth, and he was followed in an intensive care unit and fed with a baby bottle for 20 days after birth (Figure 1). He was discharged with oral feeding. In the 10th month of life, he was admitted to the Hacettepe University Hospital Emergency Room with respiratory problems and weight loss. He was diagnosed with aspiration pneumonia and was hospitalized in the intensive care unit. They started non-oral feeding and intravenous antibiotic therapy. After antibiotic therapy was completed, he was referred to Hacettepe University Hospital, Department of Otorhinolaryngology Units for a detailed swallowing evaluation. Fiberoptic endoscopic swallowing evaluation (FEES) was performed, and the absence of the epiglottis was revealed (Figure 2). Due to the uncomfortable feeling during FEES, videofluoroscopic swallowing evaluation (VFSE) was also performed with liquid and pudding consistencies for a more detailed swallowing evaluation. The penetration aspiration severity was determined by the Penetration Aspiration Scale (PAS), which is an ordinal scale consisting of eight scores from 1 to 8. A score of 1 means that the food passes safely to the digestive tract (no aspiration), scores from 2 to 5 are considered as penetration, and scores from 6 to 8 are considered as aspiration (9). The presence of nasal regurgitation was scored as "present" or "absent". The PAS score of our patient was 8, which means silent aspiration (aspiration without cough reflex). Non-oral feeding was suggested to be continued according to the swallowing evaluation. He was discharged with a nasogastric feeding tube, and a swallowing rehabilitation program was planned. His parents were also trained before hospital discharge. The therapy program consisted of thermal tactile stimulation to trigger swallowing reflex, laryngeal mobilization to support hyolaryngeal elevation, and neuromuscular electrical stimulation to activate the anterior neck muscles. The therapy program was performed five times a week over a period of 2 months by a swallowing therapist in our clinic. After 2 months, the VFSE was performed again. Penetration was detected with liquid consistency, but there was no penetration or aspiration seen with pudding consistency. The PAS scores for liquid and pudding consistencies were determined to be 5 and 1, respectively (Table 1). He had no respiratory problems, and he put on weight after the rehabilitation program (Table 2). He started a liquid-restricted oral intake. Written informed consent was received from the parents of the child.

DISCUSSION

The epiglottis develops from the hypobranchial arc at approximately the 5th week of gestational life (7). Retarded development of the hypobranchial arc at any time before the 5th week of gestational life might cause congenital deficiency of the epiglottis, including epiglottic hypoplasia or absence of the epiglottis. The epiglottis is one of the most important structures for airway protection, and the movement of the epiglottis with hyolaryngeal elevation and anterior

tilt helps to prevent aspiration during swallowing. When there is no epiglottis, arytenoid cartilages that rotate to close the rima glottis prevent aspiration.

Studies on the absence of the epiglottis are limited. Guven et al. (8) reported death due to lung infection and cardiac arrest in a 37 months male infant born with a weight of 2700 g and the absence of epiglottis. In a study conducted by Constantindes (4), tracheostomy, and gastrostomy tube were inserted immediately after recognizing the absence of the epiglottis, but exitus occurred after a few weeks due to tracheostomy obstruction. Bonilla et al.(6) followed up a 14-year-old female girl from birth, and no aspiration pneumonia symptoms were seen in that case. They indicated that sleep apnea was the most important problem in that case. The case reports showed that swallowing problems are seen in this population, and treatment methods for the feeding difficulties were reported as feeding techniques, modification of the nipple for bottle feeding, and the use of tube feeding (orogastric or nasogastric tube). However, those studies did not include any reliable or validated measurements of swallowing problems and did not describe any of the outcomes of swallowing rehabilitation in patients with absence of the epiglottis.

In our study, objective swallowing test results were used to define the swallowing problem of our case. A swallowing rehabilitation program was planned according to the swallowing evaluation. Our case started to take pudding consistency orally after two months of the rehabilitation program. Pulmonary problems were not seen, and he put on weight after the rehabilitation program. These outcomes suggest that objective evaluation of swallowing function is important to determine the appropriate rehabilitative approaches, and an effective rehabilitation program might improve swallowing function in these patients.

CONCLUSION

Morbidity and mortality rates increase due to the swallowing and respiratory problems in patients with congenital absence of the epiglottis. Because respiratory problems might lead death depending on disease severity, early detection, and management of swallowing problems are very important in these cases.

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

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REFERENCES

1. Li SP, Fang TJ, Lee SW, Li HY. A rudimentary epiglottis associated with Pierre Robin sequence. Int J Oral Maxillofac Surg 2006; 7: 668-70.
2. Holinger PH, Brown WT. Congenital webs, cysts, laryngoceles and other anomalies of the larynx. Ann Otol Rhinol Laryngol 1967; 4: 744-52.
3. Logemann JA. The evaluation and treatment of swallowing disorders. Curr Opin Otolaryngol Head Neck Surg 1998; 6: 395-400. [Crossref]
4. Constantlinides C, Cywes S. Complete median and aplasia of the. S Afr Med J 1983; 64: 293-4.
5. Koempel JA, Holinger LD. Congenital absence of the epiglottis. Int J Pediatr Otorhinolaryngol 1998; 3: 237-41.
6. Bonilla JA, Pizzuto MP, Brodsky LS. Aplasia of the epiglottis: a rare congenital anomaly. Ear Nose Throat J 1998; 1: 51-5.
7. Senggen E, Laswed T, Meuwly JY, Maestre LA, Jaques B, Meuli R, et al. First and second branchial arch syndromes: multimodality approach. Pediatr Radiol 2011; 5: 549-61. [Crossref]
8. Guven DG, Senses DA, Subasi B, Yaman H. Absence of the epiglottis in an infant with Pierre Robin sequence. Int J Pediatr Otorhinolaryngol 2011; 3: 137-9. [Crossref]
9. Rosenbek JC, Robbins JA, Roecker EB, Coyle JL, Wood JL. A penetration-aspiration scale. Dysphagia 1996; 11: 93-8. [Crossref]