

Case Report

Pyloric Stenosis in Treacher Collins Syndrome: A Case Report

Treacher Collins Sendromunda Pilor Stenozu: Olgu Sunumu

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ABSTRACT

Treacher Collins syndrome (TCS) is an autosomal dominant craniofacial developmental disorder with diverse phenotypic expression. It has a distinctive facial appearance that allows it to be easily identified. We present a case report of TCS with pyloric stenosis.

Keywords: Treacher Collins syndrome, pyloric stenosis, maxillomandibular fusion, tracheotomy

ÖZET

Treacher Collins sendromu (TCS), çeşitli fenotipik ifadeye sahip otozomal dominant bir kraniyofasiyal gelişim bozukluğudur. Kolayca tanınmasını sağlayan ayırt edici bir yüz görünümüne sahiptir. Pilor stenozlu bir TCS olgusunu sunuyoruz.

Keywords: Treacher Collins Sendromu, pilor stenozu, maksillomandibüler füzyon

INTRODUCTION

Treacher Collins syndrome (TCS) is a rare autosomal dominant mandibulofacial disease that affects 1 in 10,000–50,000 people. TCS is usually diagnosed postnatally. It is confirmed by radiographic and molecular evaluations (1). The main characteristic clinical features of the disease include bimaxillary micrognathia and retrognathia, coloboma of the lower eyelids, aplasia or microtia of the outer ear, drooping palpebral fissures and hypoplasia of the orbit, wide or protruding nose and hypoplasia of the zygomatic bone, partial or complete absence of paranasal sinuses, hearing loss, cleft palate, and choanal atresia or stenosis. Such phenotypic

features may help physicians with differential diagnoses (2,3). In addition, malformations may occur in the heart, kidney, spine, and extremities (4). Early symptoms of the disease include respiratory problems and feeding difficulties (5).

In this case, TCS was diagnosed and accompanied by pyloric stenosis. This study aimed to emphasize the difficulties encountered in airway management and provide surgical solutions to these problems.

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CASE REPORT

The mother (27 years old) and father of the male baby born at the 39th gestational week (weighing 2810 g) were second-degree cousins, and a sibling had a history of epilepsy. The physical examination revealed the following: maxillomandibular fusion, hypospadias, inguinal hernia, sacral dimple, bilateral megalocornea, left lower palpebral coloboma, and respiratory distress at baseline (Figure 1). His lips had a gap of less than 1 cm on the right. This opening was associated with the oral cavity and allowed passage of the feeding tube. Laboratory examinations were unremarkable. The midline oral cavity was not observed in three-dimensional cranial CT imaging. Irregular bone fusions were detected in the maxilla and mandible (Figure 2).



Figure 1. Singnathia, megalocornea and left palpebral coloboma with an opening less than 1 cm on the right side of the mouth only

Chromosome analysis from peripheral blood was 46 XY. A TCOF1 gene mutation was detected in diagnostic mutation genetic studies. TCS was diagnosed based on the patient's phenotypic features, radiological images, and gene analysis.

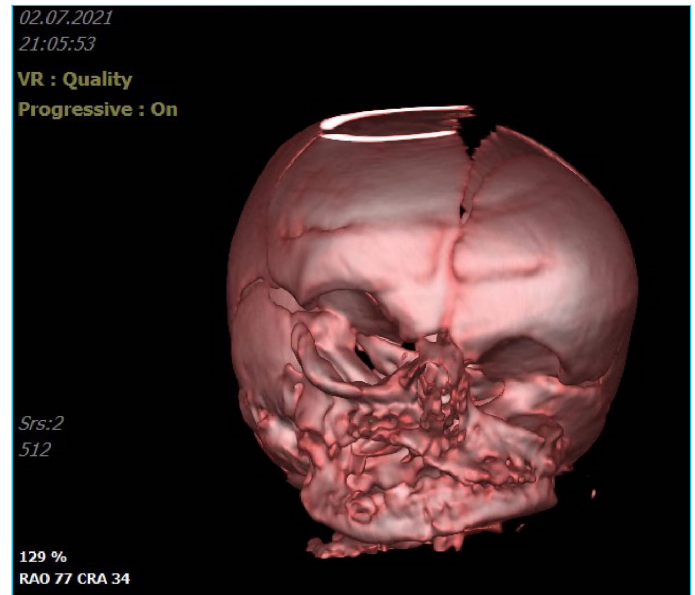


Figure 2. 3D cranial CT

On the 22nd day of the follow-up, she had vomiting without bile after projectile feeding. Pyloric stenosis was detected upon ultrasound imaging. Surgical intervention was planned for the patient owing to feeding difficulties. As airway intubation was not possible, tracheotomy was not performed. Ramstedt pyloromyotomy surgery was performed for pyloric stenosis.

DISCUSSION

TCS exhibits autosomal dominant inheritance with variable penetration. It is caused by a mutation of the TCOF1 gene, which displays linkage to the human chromosome 5q32 locus. More than 60% of TCS cases have no family history and result from a *de novo* mutation, which may be inherited from the parents in 40% of the cases (6,7). Our case also showed that 40% of the cases had a positive family history, suggesting familial mutation transfer in the TCOF1 gene.

Diagnostic features of TCS include abnormalities in the eyes, ears, nose/mouth, and facial bones. Based on these clinical features, five different clinical forms of TCS have been identified by Franceschetti and Klein: the complete form (having all known features), an incomplete form (presenting with less severe ear, eye, zygoma, and mandibular abnormalities), the abortive form (only the lower lid pseudo coloboma and zygoma hypoplasia are present), the unilateral form (anomalies limited to only one side of the face), and the atypical form (combined with other abnormalities not usually part of this syndrome) (8). In our case, the patient presented an incomplete form of this syndrome. In addition to this form, pyloric stenosis was present.

TCS has no cure. Therapy is tailored to the specific needs of each individual (5). Ramstedt pyloromyotomy surgery for pyloric stenosis was performed on our patient (due to her feeding difficulties) because intubation was not possible by performing a tracheotomy. Such patients require a multidisciplinary approach that includes a craniofacial team of pediatricians, pediatric surgeons, pediatric otolaryngologists, audiologists, plastic surgeons, geneticists, psychologists, radiologists, dental surgeons, and other healthcare professionals. However, genetic counseling should be recommended for affected individuals and their families.

CONCLUSION

TCS is an autosomal dominant craniofacial developmental disorder presenting with unusual clinical features associated with abnormalities of structures derived from the first and second branchial arches, including antimongoloid slant of palpebral fissures, lower eyelid colobomas, eyelash malformations, deafness, and facial hypoplasia. In addition to these malformations, it should be noted that patients with TCS may also have pyloric stenosis, which causes feeding difficulties. However, maxillomandibular bone fusions, which make nasotracheal intubation or tracheotomy for airway control difficult, can also be a significant issue in the context of pyloric stenosis, which necessitates surgery.

Patient Consent Form / Hasta Onam Formu

The parents' of this patient consent was obtained for this study.

Conflict of Interest / Çıkar Çatışması

The authors declared no conflicts of interest with respect to authorship and/or publication of the article.

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