



# Ketone Utilization Disorder and Hypodontia

## Keton Metabolizması Bozukluğu ve Hipodonti

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### ÖZET

Hypodontia is defined as the congenital missing of one or more teeth in one or both dentition periods. In this case report, ketone utilization disorder with oro-dental findings was reported which was previously not reported in the literature. It was concluded that dental examination is an important parameter which must be taken into consideration in the future case reports.

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**Key words:** Hypodontia, ketone utilization disorder, congenital

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### ABSTRACT

Hypodontia, bir veya daha fazla dişin, her iki dişlenme döneminde de gözlenebilecek, doğumsal eksikliği olarak tanımlanmaktadır. Bu çalışmada, literatürde keton metabolizması bozukluğunda daha önce literatürde belirtilmemiş ağız bulguları değerlendirmesi sunulmuştur. Dental muayenenin, gelecekteki olgu sunularında, dikkate alınması gereken önemli bir parametre olduğu belirtilmiştir. *The Journal of Pediatric Research 2015;2(2):99-101*

**Anahtar kelimeler:** Hipodonti, beta ketotiyolaz enzim eksikliği, kongenital

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## Introduction

Beta-ketothiolase deficiency, is a deficiency in mitochondrial acetoacetyl-CoA thiolase which is a disorder of ketone body utilization (1,2). The clinical expression of ketone utilization disorder (KUD), is highly variable. Symptoms include an asymptomatic course or intermittent ketoacidotic attacks that characterize the disease. These episodes can sometimes lead to coma. Ketoacidotic attacks are frequently triggered by systemic infections, periods of fasting, or high protein ingestion. If neurologically intact, patients experience no other issues between episodes (3-5).

Patients who lack the acetoacetyl-CoA thiolase enzyme are unable to break down proteins. This causes a toxic build-up in body tissues. Only 50 cases of this deficiency have been reported in the literature, but these studies did not provide evidence of orodental findings. This case report presents a child patient with hypodontia and KUD.

## Case Report

A 10-year-old male patient was referred to the Department of Pediatric Dentistry, Faculty of Dentistry, at Gazi University for general dental care. His appearance was different from

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other children of his age; in particular, he was shorter in height. The patient presented with a history of KUD. Additional patient records were subsequently procured. Informed consent was obtained from the parents of the children.

Our patient had been diagnosed with his first ketoacidotic episode as a 9-month-old at the Department of Pediatric Metabolism and Nutrition, Faculty of Medicine, at Gazi University. He was brought to the infant emergency clinic due to high fever, cough, and tachypnea. In the course of evaluation at the clinic, a metabolic analysis was ordered due to severe metabolic acidosis and a diagnosis of blood ketones found in the initial examination. An increase in the levels of acylcarnitine and 3-OH isovalerylcarnitine was observed in an analysis with blood spot electrospray tandem mass spectrometry and an acylcarnitine analysis. Moreover, the urinary organic acid analysis showed a significant increase in 2-methyl-3-hydroxybutyric acid tiglylglycine. He was discharged from the hospital after being placed on a low-protein diet. He suffered no additional attacks after that time and continues to be monitored.

The dental examination showed normal intraoral soft tissues. An intraoral examination revealed the presence of primary maxillar laterals and primary mandibular central incisors. Considering the patient's age, the eruptions of the anterior mandibular permanent teeth, the permanent mandibular left incisor tooth, and the permanent maxillar right and left lateral teeth were regarded as delayed (Figure 1). Also deep carious lesions in the primary molar teeth were detected during the clinical observation. However, the patient had maintained good oral health habit and was motivated to prevent decay. On the other hand, seven primary molar teeth and one permanent molar tooth had dental caries.

Radiological evaluation revealed five congenitally missing permanent teeth, thereby indicating severe hypodontia (Figure 2). The patient had no family history of delayed eruption or hypodontia.

Dental restorations were administered, the patient was planned to be monitored until the end of his craniofacial growth. An individualized prevention program including fluoride and sealant applications besides fluoride containing toothpaste and mouth rinses has been provided at both the dental visit and through home care.

Congenitally missing permanent teeth was an important dental observation in this case as no previous studies have reported the association of hypodontia and KUD.

## Discussion

In the dental examination for this case, five congenitally missing permanent teeth were diagnosed radiographically. At the age of ten, usually all permanent teeth, including the third molars, are visible on a radiograph. Moreover, all permanent teeth except the third molars have typically erupted by the age of 12 to 14. A tooth is defined as congenitally

missing if it has not erupted in the oral cavity and not visible on a radiograph within its eruption period. Hypodontia is characterized by the partial or total, congenital lack of formation of one or more teeth on one or both dentitions. Heredity is the main etiological factor in hypodontia, and the principal clinical features include reduction in the number, size, and form of the teeth and late eruption. Our patient had no family history of delayed teeth eruption or agenesis. Despite this lack of family history, our patient was congenitally missing his mandibular central teeth and maxillar right lateral teeth. According to Nunn et al., the absence of mandibular incisors is very rare (6). On the other hand, mandibular lateral incisor agenesis has a higher prevalence rate in the Japanese population (7). Its prevalence in other populations is about 0.5% (8). Also, the patient's maxillar second molars were not radiographically visible, but the mandibular the second molars were coronally formed. However rudimentary development of molars in such cases should not be ignored (9).



Figure 1. Intraoral dental examination



Figure 2. Panoramic radiographs shows missing teeth

Literature research showed that there was a lack of dental examination of KUD patients. Although the case described herein may be exceptional, to pay attention to oral findings is very important should ignored.

As described in the introduction, ketoacidotic attacks are frequently triggered by systemic infections, periods of fasting, or high protein ingestion (5). Symptoms include intermittent episodes of severe metabolic acidosis and ketosis accompanied by vomiting, diarrhea, and coma that can progress to death. Great clinical heterogeneity is observed among patients (4). This 10 year old KUD patient had no ketoacidotic attack history after his first ketoacidotic episode at 9 months of age.

The effects of attacks on dental management are not clear. No specific dental management protocol has been determined, but dentists must be prepared to manage medical emergencies that may arise in practice. If an attack occurs during a dental procedure, maintaining the patient's airway, providing basic life support activity, and escorting the patient to the hospital should be priority action in order to avoid any clinical consequences.

Our patient has a meal plan that meets his unique needs for growth and development. He just started to drink milk about 6 months ago in order to increase his calcium intake due to initial osteoporosis symptoms. Clearly, calcium is a major nutrient for tooth health as well. Nutrition and oral health are inextricably linked. Dietary calcium and phosphorus in the form of phosphates are incorporated locally into the microbial plaque, thereby reducing demineralization and enhance remineralization. Dairy foods are the richest dietary source of calcium. Studies evidence the protective effect of milk and milk products, such as cheese (10). However, our patient may not benefit from the increased intake of dairy foods. Calcium deficiency from the special diet of the patient may also explain the increase in dental caries. Inadequate nutrition can also effect the formation of tooth enamel. Abnormal formation of tooth enamel leaves some areas more susceptible to decay. Dental risks may be reduced with the mechanical/chemical removal of plaque (i.e., good oral hygiene), the chemical (i.e., antimicrobial) modification

of plaque, the use of fluorides, dietary modifications, and salivary stimulation, all of which help to better control the underlying disease (11-13).

More evidence is needed to determine whether KUD and hypodontia are truly related. Therefore, we suggest that dental examination is an important parameter that must be taken into consideration in future case reports.

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