Introduction: Early tooth loss could be the consequence of the local or systemic diseases. We presented an odontohypophosphatasia case and family investigation with otosomal dominant mutation in ALPL (TNSALP).

Material and Methods: A three-year-old boy admitted to our pediatric endocrinology clinic with premature exfoliation of anterior incisors and canines without any other dental or gingival disease. His loss of teeth had begun thirteen months ago. Parents were first degree cousins. There were no any health problems in his 7-year-old brother and 14-month-old sister. Also his parents had no clinical symptoms. In his physical examination; his height was 97.8cm (10 p.), his weight was 14.5 kg (10 p.). There were no pathological sign in his physical examination without toothloss. In his biochemical analysis; Ca:9.7 mg/dl, P:5.9 mg/dl, ALP:70 U/L, PTH: 32.2 pg/ml 25hydroxy Vitamin D: 18.9 ng/ml. We considered that the patient have odontohypophosphatasia. We performed ALPL gene analysis. PCR techniques were used to amplify the all translated exons of the ALPL gene. Sanger sequencing technique was used for mutation analysis and ALPL gene analyzed with ABI 3130 Sequencer device. Heterozygous otosomal dominant c.346G>A (p.A116T) mutation was detected in fifth exon of ALPL. ALPL gene analysis was performed to all members of the family. While his father has no mutation, his mother, brother and sister have the same heterozygous mutation in the same locus.

Discussion and Conclusion: Hypophosphatasia manifests itself principally by faulty mineralization of the bones and teeth, and is caused by defects in the tissue non-specific alkaline phosphatase (TNSALP) gene on chromosome 1 (1). The predominant mode of inheritance is autosomal recessive, although about 10% show a dominant pattern. The symptoms are highly variable in their clinical expression, which ranges from stillbirth without mineralized bone to early loss of teeth without bone symptoms. People with dominant hypophosphatasia usually experience moderate symptoms, such as the premature exfoliation of fully rooted primary teeth (2).

Odontohypophosphatasia should be included as a differential diagnosis in children with early loss of primary teeth. It can be presented without extremely low alkaline phosphatase levels.

References: