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VACTERL SYNDROME: DIFFERENT DENTAL FINDINGS

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CASE REPORT

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ABSTRACT

VACTERL syndrome is a congenital malformation that effects multisystem. The characteristic symptoms of this syndrome are vertebral anomalies (V), anal atresia (A), cardiac defects (C), tracheoesophageal fistula together with esophageal atresia (TE), radial dysplasia, renal anomalies (R), and limb defects (L). The prevalence of VACTERL syndrome is 1/10,000-1/40,000. Clinical diagnosis can be made with at least three out of the symptoms. Oral manifestation of VACTERL syndrome has not been reported in the literature. Oral findings are the most noticeable point of his syndrome. So, we aimed to draw attention to oral findings in the syndrome in this study. This is the first case report that investigated oral/dental findings which accompany supernumerary teeth and early tooth eruption as well as medical findings.

INTRODUCTION

Quan and Smith described VATER syndrome in 1972 that showed vertebral anomalies (V), anal atresia (A), tracheoesophageal fistula together with esophageal atresia (TE), and radial dysplasia (R). In 1973, renal abnormalities (R) were added to the description by the same researchers,[1] and in 1974, the definition of VATER syndrome was changed as VACTERL syndrome by the addition of Temtamy and Miller's cardiac (C) and limb (L) defects.[1] Clinical diagnosis was made due to the presence of three of full VACTERL symptoms.[1]

The prevalence of VACTERL syndrome is 1/10,000 -1/40,000.[3] Non-genetic risk factors, such as maternal diabetes may influence the etiologic possibility as well as the precise reason of the syndrome has not been defined yet.[4] A high percentage of the diagnostic criteria is observed in consanguineous parents or the twin brother/sister of individuals diagnosed with VACTERL syndrome.[5] Chromosomal aberrations, single gene mutations,[5] and microdeletions of the Xq25 region[3] are also reported in VACTERL syndrome.

Oral manifestation of the syndrome has not been studied too much in the previous reports. This is the first study in the literature with oral/dental findings and that defined the presence of supernumerary teeth and early tooth eruption as well as medical findings.

CASE REPORT

A 5-year-old male patient was referred to us by outside clinics due to the inability of examinations and

treatment in normal clinical situations. The patient had diagnosed with VACTERL syndrome four years ago. Diabetic (type II) mother had six pregnancies and resulted with four surviving infants and two stillbirths. The two children whose hands had six fingers, died immediately after the birth. Our patient was born in 31 weeks at 1,700 g weight via caesarean section and was the sixth child of the consanguineous parents. The patient had congenital scoliosis and underwent an operation for a urethra pelvic junction obstruction, non-functional left kidney, and Morgagni hernia and also anal atresia operation at 3 days old that controlled in intensive care unit for 22 days. There were improvements in his congenital atrial septal defect and patent foramen ovale, which was observed at 2 years old. He has congenital hip dysplasia, and his right extremity was 2 cm shorter than the left. However, the patient was able to walking. But, the patient with insufficient cognitive improvement is unable to speak and had been using hypothyroidism drug at three years and eight months of age. The patient has also bilateral hand polydactyly, prominent frontal regions, a flattened nasal bridge, and slant-eyed (Fig. 1). However, there are not any temporomandibular joint problems. The patient is 35 kg weight and 115 cm height at the last record, which showed that he was overweight according to the 2004 World Health Organization criteria. Frequent snack and fruit consumption times were followed for three days the diet analysis of the patient.

Patient completed a consent form and approved the treatment. We observed that the patient had mouth breathing, higher saliva viscosity and buffering capacity, and also analyzed that salivary pH was 7.6, and presented a positive *Streptococcus mutans* culture. All of the deciduous teeth are at the radix levels even though the 7.6 pH. But, the possible reason may be higher buffering capacity, poor oral hygiene and specially feeding with carbohydrate. Unfortunately, tooth brushing habit of the child was not good and the parents did not care about the importance of oral hygiene among his medical problems.

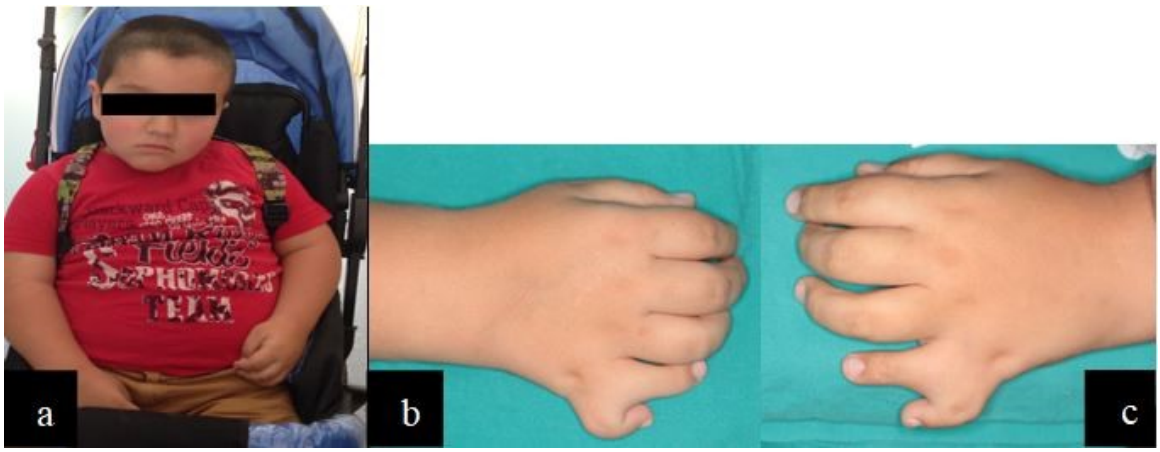

Figure 1 Physical aspects

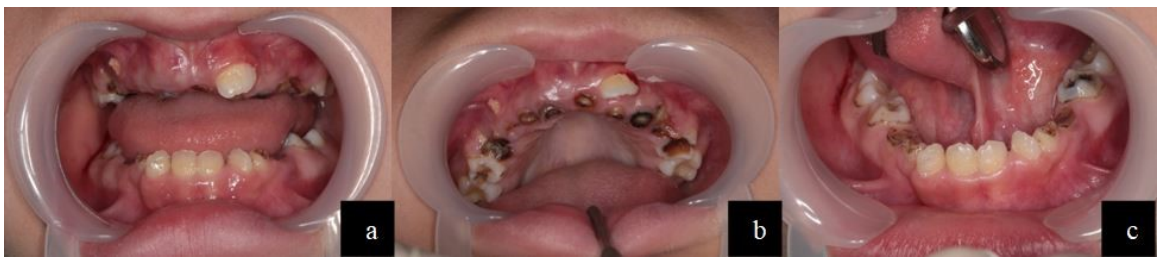

Figure 2 Intraoral clinical aspects of dental condition

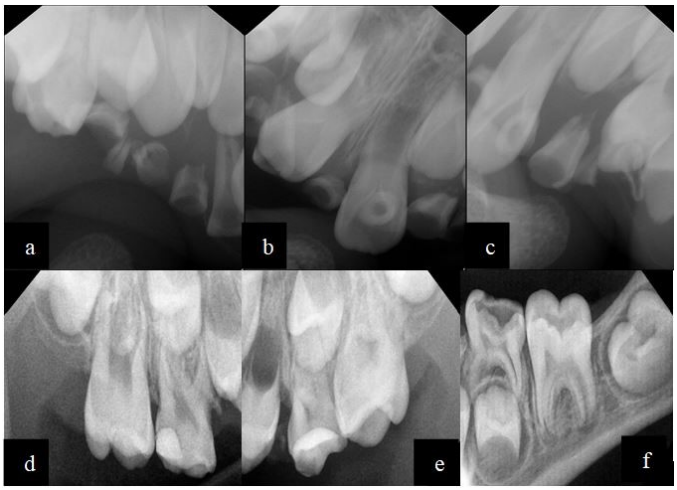

Figure 3 Radiographic view

The oral mucosa, frenulum, tongue, and gingiva were determined to be normal upon intraoral examination. General black extrinsic colorations were present on the teeth surfaces. All of the first permanent molars and teeth numbers 21, 32, 31, 41, and 42 were erupted immediately (Fig. 2). Supernumerary teeth in the right maxilla region (placed between the 11-13 numbered teeth) were examined in the periapical radiographs. Teeth numbers 55 and 65 had dentinal caries, and all of the other deciduous teeth were at the root levels except 85 numbered tooth (Fig. 3). Teeth, 55 and 65, were restored with compomer filling material, and all of the deciduous teeth radixes, except 85, were extracted under general anesthesia. A conservative program, including dietary suggestions, oral hygiene motivation, fluoride applications, and a maintenance program for checking supernumerary teeth, was planned for the patient's age and oral pathologic eruption. The parents were motivated to schedule regular checkups.

DISCUSSION

VACTERL syndrome is a congenital malformation condition that effects multiple systems. There is no standard approach for the diagnosis.[6] Many similar clinical findings with Vacteryl syndrome have been described. Therefore, effective and certain differential diagnosis should be done. For example, CHARGE syndrome is distinguished by congenital ophthalmic anomalies and choanal atresia; Fanconi anemia demonstrates remarkably similar phenotypic characteristics but differs in hematologic abnormalities and pigmentary changes; Feingold syndrome shows some different properties, such as shortened second and fifth digits and microcephaly; Di George syndrome differs from VACTERL syndrome in thymic abnormalities, conotruncal cardiac anomalies, facial dimorphism, and hypocalcemia.[7] There are no differences in the oral mucosa and surrounding tissues upon oral examinations. Previous studies defined increased salivary flow,[4] fusion,[8] and single maxillary central incisors.[9] However, there are not explanations for supernumerary teeth presentation. The patient is not able to use a space maintainer because of the inadequate cognitive development. So, a regular checkup program is organized for the patient.

Preterm birth, weight of 1,700 g, and hypothyroidism stimulated early tooth eruption. Starting the completing of the root apex of the first permanent tooth and the clinical occlusal relationship were uncommon features. These features are confirmed by periapical radiographs, because of the difficulty of panoramic radiographs. Bone resorption related to the bad oral hygiene, obesity and hypothyroid drug caused early tooth eruption. Adverse effects of hypothyroid drug may influence the eruption times, and hyperthyroidism symptoms

can be observed in patients depending on irregular hypothyroid drug intake as defined in the study of Franklyn and Boelaert[10]. However, the difficulty of panoramic radiograph taking complicates the evaluation of eruption of teeth.

In conclusion, VACTERL syndrome should be evaluated in terms of oral findings, which are among the diagnostic criteria of many syndromes. However, the number of studies on this subject is very limited in the literature. Early diagnosis of the syndrome can minimize the oral findings of the syndrome. Consultation of the patient to a pediatric dentistry in time may be very important factor for the patient and the treatment.

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