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DENTAL MANIFESTATIONS IN A FEMALE PATIENT WITH APERT'S SYNDROME

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Abstract: Viral Apert's syndrome (AS) is a rare congenital disorder with autosomal dominant inheritance and is characterized by irregular craniosynostosis, syndactylia of hands and feet, mid-face hypoplasia, hypertelorism and anomalies of central nervous system, heart and kidneys. AS has been associated with mutations in Fibroblast growth factor receptor 2 (FGFR2) located on chromosome 10q (10q26). Dental anomalies are common in AS. We report on a 6-year-old AS patient with complex dental anomalies. A 6 year-old female patient with AS was presented to the dental clinic with complaints of teeth decay and embedded teeth. She had dysmorphic facial symptoms including mid-face hypoplasia, low-set ears, hypertelorism, prognathic mandible, steep wide forehead, down-slanting lateral canthi and palpebral fissures. She had syndactyly of third and fourth digits of both hands. Arachnoidal cyst was diagnosed previously. She had intellectual disability. Radiography showed that there were more than one embedded teeth. Upper first premolar and canine teeth were displaced. She had teeth agenesis of the maxillary lateral incisor. Her maxilla and mandible were narrow. The maxillary dental arch was v-shaped. Orthodontic treatment was planned for the future because the patient was too young. The aim of the present report is to show the dental manifestations in case with AS. The treatment and management of AS require a multidisciplinary approach.

Keywords: Apert's syndrome, Dental anomalies, Orthodontic treatment.

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1. Introduction

Apert's syndrome (AS) (acrocephalosyndactylia type I) (OMIM #101200) is a rare congenital disorder that causes severe craniosynostosis (premature fusion of cranial sutures), craniofacial dysmorphic features, symmetrical syndactyly of the digits, and tooth anomalies , as well as a variety of associated congenital anomalies of the brain, heart, limbs, and other organ systems (Fadda et al., 2015). The prevalence of AS is 1 in 65,000 to 160,000 live births (Park et al., 1995, Cohen et al., 1992). AS is caused by mutations in Fibroblast Growth Factor Receptor 2 (FGFR2) located on the long arm of chromosome 10q (10q 25-26). This disorder is associated with autosomal dominant inheritance but most of the cases are sporadic (Gupta et al., 2013). More

than 98% of AS cases are caused by FGFR2 de novo mutations. Dental anomalies are very common in AS. The present paper describes the dental manifestations in a 6-year-old girl previously diagnosed with AS.

2. Case Report

A 6 year-old female patient diagnosed with AS presented to our department with the complaints of embedded teeth and teeth decay. Extra-oral examination, intra-oral examination, and radiographic evaluations were performed after detailed medical history of the patient was taken. She was born at term via a vaginal delivery after an uncomplicated pregnancy. She was the daughter of a nonconsanguineous parents as the the 3rd living child from the the 3rd pregnancy. No other family members had the same health conditions. Arachnoidal cyst detected previously by CT imaging. She had intellectual disability. Her speech was limited to a few words. She was not able to urinate. She had hyponasal resonance. Her medical history revealed that she had undergone plastic surgery for the treatment of syndactyly three years ago. Clinical examination showed that the girl had a flattened occiput, short anteroposterior diameter with high, full forehead, craniofacial asymmetry, low-set ears, mid-face hypoplasia, depressed nasal bridge, hypertelorism, down-slanting palpebral fissures, prognathic mandible, steep wide forehead and syndactyly of third and fourth digits of both hands (Figure1 and 2).

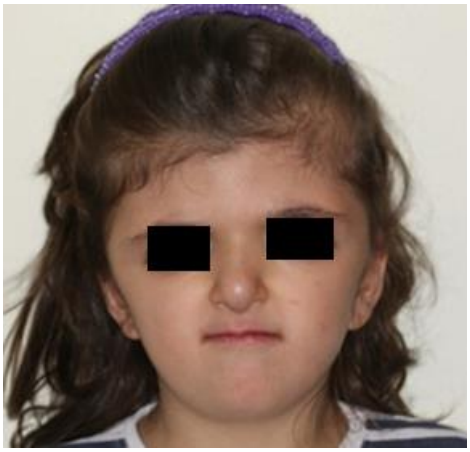

Figure 1. A facial photograph showing depressed nasal bridge, hypertelorism, and downward slanting outer canthus of eyes.

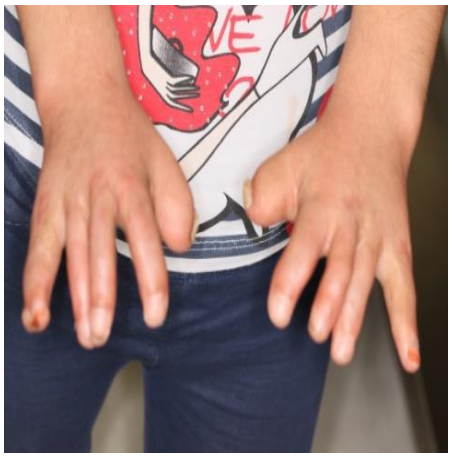

Figure 2. Bilateral symmetrical syndactylia of fingers.

Intraoral examination showed a high arched (V-shaped) palatal vault, teeth decay and embedded teeth (Figure 3).

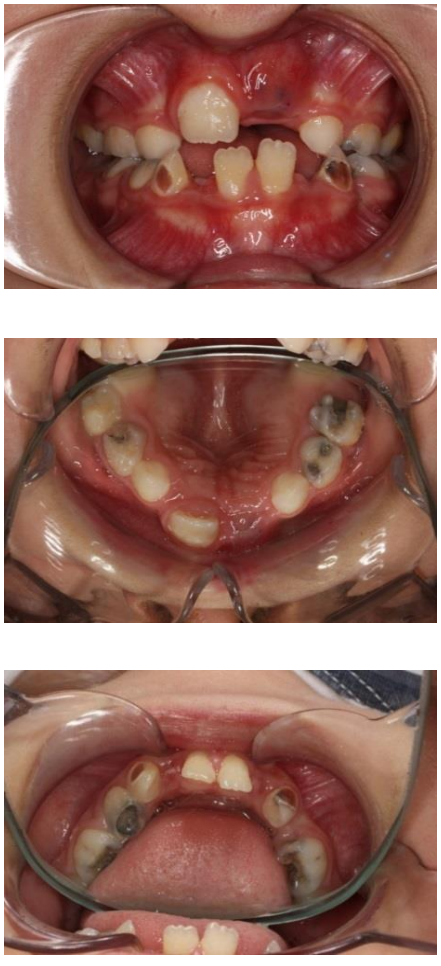

Figure 3. Intra oral views.

The panoramic radiograph revealed retention of the primary teeth (mandibulary second incisor teeth).

Maxillary canine and first premolar teeth were displaced. She had tooth agenesis of the left maxillary lateral incisor (Figure 4).

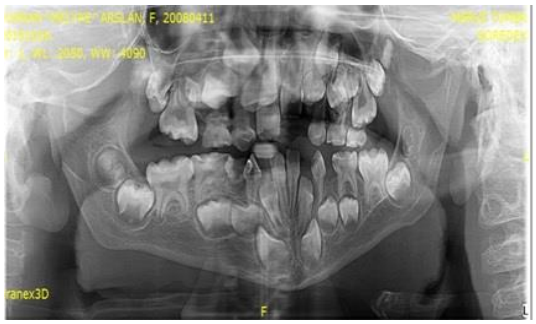

Figure 4. Panoramic radiography of the case.

Her maxilla and mandible were narrow. The maxillary dental arch was v-shaped. Supernumerary teeth and cleft palate were not present. Teeth decays were treated by a pediatric dentist. Orthodontic treatment was planned in the future because the patient was underage and we were informed that orthognathic surgery may be required.

3. Discussion

AS is an extremely rare developmental genetic disorder which first reported by Whearon in 1894 and then reviewed by Eugene Apert in 1906 (Fadda et al., 2015). AS, one of the most severe craniosynostosis syndromes, is characterized by irregular craniosynostosis, symmetrical syndactylia of hands and feet, mid-face hypoplasia, hypertelorism and anomalies of central nervous system, heart and kidneys. Although AS has a dominant inheritance pattern, most of the cases are sporadic and exhibit paternal effect (Kumar et al., 2014). FGFR2, responsible for AS, is one of the four Fibroblast Growth Factor Receptors involved in bones formation. Approximately 99% of the cases with AS had two missense mutations that involve C-to-G transversions at adjacent codons in exon IIIa of the gene (Ser252Trp or Pro253Arg; Ibarra-Arce et al., 20156). All these FGFR2 mutations cause a gain-of-function mechanism and the development of abnormal cranial suture, which finally induce craniosynostosis (Fadda et al., 2015).

Our patient was previously diagnosed with AS and represented typical facial appearance of the syndrome. Central nervous system (CNS) abnormalities in AS were reported in various studies. Among these anomalies are the corpus callosum anomalies, hypoplasia or absence of the septum pellucidum, cerebral cortex dysplasia, pyramidal tract abnormalities, and hypoplasia of septum pellucidum, cerebral cortex dysplasia, heterotopic gray matter, encephalocoele, and megalencephaly (Cohen and Kreiborg, 1990; Bhatia et al., 2013). Aracnoidal cyst was detected in the patient previously. A significant number of patients with AS suffer from mental retardation (Patton et al., 1988) and this may be partly due to CNS anomalies. Our patient had also intellectual and speech disability.

Furthermore, orodental anomalies are seen frequently in AS patient. About 75% of AS patients were reported to have cleft palate or bifid uvula (Kreiborg and Cohen, 1992). Affected individuals exhibit other dental malformations including supernumerary teeth, thick gingiva, impacted teeth, ectopic eruption, and delayed eruption. Tooth agenesis may be seen in AS patients (Letra et al., 2007). Cleft palate, bifid uvula and supernumerary teeth were not present in our patient, but she had maxillary lateral incisors agenesis, delayed eruption and embedded teeth. Teeth decays were treated by a pediatric dentist, and orthodontic treatment was planned for the future as the patient was underage.

The morphological defects in AS patients are likely to obstruct physiological functions. In these patients, ensuring oral hygiene is of particular importance since they mostly have mental retardation and hand deformities. The use of electric tooth brushes and fluoride mouth rinses may be beneficial in these patients.

Although there is not specific treatment for AS, there is much to do to prevent or treat complications and to help children grow under as normal conditions as possible. As it is the case in many syndromic cases, the treatment and management of these patients require a multidisciplinary approach. Because dental symptoms are common, orthodontists can contribute to the improvement of a patient's appearance and functioning of dentoskeletal structures by combining orthodontic and orthognathic surgical treatment.

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