Adams-Oliver Syndrome: a Case Report

SUMMARY

Background: Adams-Oliver Syndrome has been described by Adams and Oliver in 1945. Original definition, along with aplasia cutis congenital syndrome and limb defects, has neurological and cardiological problems. In the first description, genetic defect passes variable autosomal dominant pattern. Afterwards the autosomal recessive and sporadic cases were published. Case Report: 11-year old male patient complained of mobile teeth admitted to our clinic. He was noted to have characteristic view with distal-phalanx and nail hypoplasia of his hand and feet with occipital scalp defect. We consulted to genetics because of these findings and learned that he has the Adams-Oliver Syndrome. The patient has some orofacial manifestations including high-narrow palate, fissured tongue, crowding, dysmorphic facial features, facial asymmetry, deep-philtrum, delayed eruption, class III malocclusion. The extraction of mobile deciduous teeth, restoration of caries and also oral hygiene motivation was made. Then, the patient was referred to orthodontics. Conclusions: Adams-Oliver syndrome represents rare congenital alteration, insufficiently documented in scientific literature. This shows the need to document news cases.

Key words: Adams-Oliver Syndrome, Genetic Diseases, Dental Findings

Introduction

Adams-Oliver syndrome (AOS) was first defined in 1945. The syndrome is characterised by aplasia cutis congenita (ACC) and terminal transverse limb defects. While its original definition was based on ACC syndrome and limb defects, neurologic and cardiac problems accompany the disease. ACC lesions are among the most classical findings of the disease and frequently are seen along the posterior sagittal suture as hairless spaces of different sizes on the skull. These lesions generally are seen on the parietal and occipital midline; however, they also can appear on the abdomen and legs. ACC lesions also can appear at birth as an epithelised scar. ACC lesions are generally bigger than 5 cm, and they tend to heal within the first month after birth. Major lesions have a high probability of including the skin and cerebral cortex. This brings the possibility of complications, such as haemorrhage and thrombosis, which may cause infection and death. Another symptom of the disease includes limb defects (partial or complete absence of all toes or fingers, which looks like amputation), and they may vary from mild (unilateral or bilateral short distal phalanx) to severe. Lower limbs are more affected than the upper limbs in almost all of the cases. Aplasia cutis congenita and terminal transverse limb defects should be researched in the differential diagnosis of the disease. Terminal transverse limb defects (TTLD) can include short distal phalanx, brachysyndactyly or ectrodactyly. Early embryonic vascular impairment is considered the underlying reason for the pathogenic mechanism. A wide spectrum of defects indicates impairment of multiple development pathways. Congenital heart diseases associated with the syndrome are seen in more than 20% of AOS patients and present a serious mortality risks. The most frequent cardiac defect resulting from anomalies in systemic vascular structures has been stated as cutis marmorata telangiectasia congenita (CMTC). A phenotypic variation seen in AOS is differentiated from an autosomal dominant feature. The low incidence of limb defects makes the disease rare and difficult to diagnose.
of the disease in relatives is compatible with autosomal recessive gene transmission. In addition, sporadic cases which present with similar clinical features show that the genes which cause the disease occur as a result of de novo mutations. Cardiovascular malformation/dysfunctions (23%), brain anomalies and, less frequently, kidney, liver and eye anomalies can be given as examples of frequently seen characteristics.

A review of the literature about the disease showed that there were a great number of studies about the genetic aspect of the disease. However, it was found that there were limited numbers of studies about the orofacial findings of the disease. The purpose of this study is to present the clinical radiological findings of a patient with AOS.

Case Report

An 11-year-old male patient was admitted to Konya Necmettin Erbakan University’s Department of Pediatric Dentistry. The patient was found to have occipital scalp defects, characteristic appearance in distal phalanges of hands and feet and nail hypoplasia. As a result of these findings, the patient was consulted to Konya Necmettin Erbakan University, Meram Faculty of Medicine, Department of Genetics. The patient was diagnosed with AOS.

The patient had some orofacial findings thought to be associated with the syndrome, including occipital skin defect, growth retardation when compared with peers, dysmorphic facial features, asymmetry of the face and deep philtrum, distal phalanges in hands and feet and nail hypoplasia (Figure 1-3). As a result of consultations with Konya Necmettin Erbakan University, Meram Faculty of Medicine, Department of Pediatrics, it was learned that the patient was under control due to his heart disease. In addition, the patient’s biological parents were non-kin, and the other siblings did not have the syndrome.
first molars. The panoramic film of the patient showed that the permanent teeth, which were still germ, were not missing (Figure 7). The treatments were conducted under general anaesthesia since the patient did not cooperate and had a strong nausea reflex.

Figure 4. Preoperative intraoral photograph of lower jaw

Figure 5. Preoperative intraoral photograph of upper jaw

Figure 6. Preoperative photography showing malocclusion and gingivitis

Figure 7. Preoperative OPG

Figure 8. Post operative anterior photography (healing of the gums after detertrage and oral hygiene motivation)

Figure 9. Post operative intraoral photograph of upper jaw

Figure 10. Post operative intraoral photograph of lower jaw

Figure 11. Post operative OPG (Six months follow up)
The patient and his family were informed about the treatment, and oral and written consent were taken. Pretreatment photos were taken in the first session. The patient was given oral-hygiene training. In the second session, all the treatments were conducted under general anaesthesia at Konya Necmettin Erbakan University, Meram Faculty of Medicine. The maxillary left first molar, maxillary left second premolar, maxillary right second premolar and mandibular right pre-canine teeth were extracted. The mandibular right second molar had a cavity, and it was restored with compomer, or composite resin (Figure 8-11). Thus, the patient’s restorative and surgical operations were completed. The patient was directed to the Department of Orthodontia for Class III malocclusion treatment. However, the treatment was delayed due to the patient’s uncooperative behaviour. The patient’s regular controls are still continuing in Konya Necmettin Erbakan University’s Department of Pediatric Dentistry.

Discussion

AOS is a rare congenital disorder consisting of distal limb anomalies (absent or short phalanges) and vascular anomalies (dilated veins, cutis marmorata and haemangiomas). Cardiac and ophthalmic anomalies, facial asymmetry, cryptorchism and spina bifida occulta are among rare symptoms. According to a literature review, there are a great number of studies about the characteristics of the disease. However, there are limited numbers of studies about the orofacial findings of the disease.

In 1945, Dr. Adams and Dr. Olivers reported eight members of a family who had ACC and distal transverse limb reduction defects. Since then, AOS has shown variations from the mildest form of aplasia cutis congenita to skull defects at birth and demode wide areas on the skin to limb reduction defects and cutis marmorata telangiectatica congenita. Mortality is low in patients with AOS, representing 20% of known patients. The most common reason for mortality is haemorrhage in aplasia areas, secondary infections and meningitis.

Dentistry is one of the important departments which require a multidisciplinary approach of AOS. The dentist can prepare a program to prevent or eliminate the changes according to the patient’s age, oral and dental health. In order to succeed, it is important to know the varying clinical findings of AOS. Hepatic neurological, vascular and cardiac anomalies have been found in 13% of AOS patients. Thus, a program should be prepared for dental treatment after suitable medical assessment. Following this, protective measures are very important in preventing endocarditis. These should include prophylactic antibiotic use in AOS patients who have congenital cardiopathy or bone defects which cause the exposure of parts of the brain.

The clinical symptoms in this case are quite varied, just like other case reports; there is a consensus on symptoms, such as presence of distended veins, limb deformities in the form of syndactyly (webbed fingers and toes) proximal and medial phalans reduction, nail hypoplasia, club foot, eye anomalies, growth hindrance, hydrocephaly, psychomotor delay, facial cleft, bi-lateral cleft lip and Grade III cleft palate. Facial cleft, bi-lateral cleft lip and Grade III cleft palate are rare symptoms which are seen in 0.3% of individuals.

The treatment was oral and dental in this study. At this stage, the objective was to increase the oral hygiene and to make the hypocalciﬁed areas in the teeth resistant to the consumption of carbohydrate-rich foods. Thus, the risk factors which caused decay formation were decreased and oral health was improved.

Conclusions

AOS is a rare congenital disorder which has not been sufficiently studied. This shows the need for more case reports. AOS requires a multidisciplinary approach for successful treatment and control. Paedodontics is a part of the working team that provides the means to develop oral and dental conditions and controls the risk factors to prevent decay development as well as harmful habits and malocclusion. AOS requires multidisciplinary assessment of protective measurements in oral and dental treatment. A special approach is necessary for the treatment of children with AOS, and thus, the responsibility of pedodontists increases twofold.

References


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