Introduction

Pfeiffer’s syndrome (PS) is a rare, autosomal dominant, variable-severity acrocephalic skull characterized by Pfeiffer in 1964, characterized by a declined midface, hands and feet, and large thumb and large foot syndactyly [10]. PS is defined as a syndrome characterized by the appearance of skull and leg anomalies in eight individuals from three quadrants [14].

This syndrome, which is seen in 100,000 cases, is usually described as a syndrome with a craniosynostosis of coronal sutures, a large hand and a toe, and a rare, autosomal, dominant transition syndrome [4, 14]. Genetic mutations are rarely possible. Coronal and lambdoid occasionally play a role in the craniosynostosis of sagittal sutures [3].

Characteristically, clinical images include; turribrachycephaly (sharp rounded head shape, pronounced and protruding forehead) and maxillary hypoplasia, exophthalmos, small zygoma and undeveloped maxilla. Sometimes partial soft tissue syndactyly can also be seen in the hand. A flat midface, wide feet and thumbs are also among the clinical features [1].

Eyes seem swollen due to shallow orbitas that is caused by maxillary hypoplasia (ocular proptosis). Hypertelorism and downward sloping palpebral fissures are common and strabismus is often seen. Intelligence is normal. In this case report, we aim to present the follow up and primary teeth extractions performed in 4 years old girl with type 1 Pfeiffer Syndrome.

Other characteristics of PS are; lower ear lobes and finger anomalies [7]. Oral findings include mandibular prognathism, malocclusion, cleft pseudo-palate and bifid uvula, enlargement of the alveolar bone, high palatal arch, and crowded teeth [3, 7]. Cohen classified PS into three clinical subtypes according to phenotype severity. Type 1 (classical) PS includes individuals with mild findings of normal neurological and mental development. Type 1 (classical) PS may be dominant. Type 2 PS, clover leaf skull, broad hands and feet, severe exophthalmos, central nervous system involvement, elbow ankylosis or synostosis. Type 3 PS, similar to Type 2. The only difference is that the clover leaf skull is not visible. Type 2 and 3 cause premature death due to severe neurological complications and various vascular problems [6, 9]. In this case report, we aimed to present a case of 4-year-old girl who had Type 1 PS in our clinic.
Case Report

As a result of physical examination performed in a 4-year-old girl who applied to our clinic due to dental problems, turribraksephaly, maxillary hypoplasia, ocular propitosis, hypertelorism, strabismus, exophthalmos were detected (Figure 1). In our hospital history, it was understood that the mother of the patient gave birth her 1.800 g weight and 48 cm height with cesarean section. Her mother has done normal pregnancy period and her mother and father are 1st degree relative marriage. Post-natal infants have a diagnosis of PS due to turribrachycephaly, maxillary hypoplasia, ocular proptosis, hypertelorism, strabismus, exophthalmos. It wasn’t found soft tissue syndactyly and wide hands and feet that seen in the PS, and the intelligence of the case was reported to be normal (Figure 2, 3).

The intaoral clinical and radiological examination revealed that oral hygiene was worse and had widespread quite a lot tooth decay and abscess (Figures. 4, 5 and 6). Temporary teeth with all ablative and wide caries defects were extracted under local anesthesia (Figures. 7 and 8) and 2% NaF gel application was performed for prophylaxis in order to prevent new caries. The case is still under control.

Discussion

Although the incidence of PS is unknown, it is reported to be 1 in 100,000 [9]. It has also been reported that cases with Type 1 PS have good prognosis. Patients with Type 2 and Type 3 prognosis have reported early death in spite of aggressive medical and surgical treatment [10]. Our case was Type 1 PS and the clinical findings were relatively acceptable in comparison with other types. The etiology of PS is not entirely clear, but it is thought to be a craniofacial syndrome and mutations in the fibroblast growth factor (FGFR) receptor. It is known that FGFR 2 and FGFR 1 receptors are caused by mutations in exons IIIa and IIIc [12]. Some studies have reported that these mutations originate from the chromosome 8p11.22-P12 in the FGFR1 gene and from the chromosome 10q25-q26 in the FGFR2 gene [11]. These mutations are thought to be related to the older ages of the parents. These mutations are associated with the fact that sperm have a selective advantage for survival, especially for men [8]. Her parents have been married to relatives of the first degree. We think the syndrome is for this reason. Bannink et al., Reported that obstructive sleep apnea developed in approximately 50% of patients with Apert, Crouzon, or PS due to moderate hypoplasia [2]. Boutros et al., reported in patients with Apert, Crouzon, and PS, a narrowing
of the cranial tendon caused narrowing of the condylar area, and mandibular anomalies appeared in the form of decreasing biconical enlargement with the enlargement of the bigonial almost normal [1, 5]. In our case, only turribrachycephaly, maxillary hypoplasia, ocular proptosis, hypertelorism, strabismus, exophthalmos has been observed and we are still following to detect possible anomalies early. Patients that referred in the dentist clinic are often healthy individuals, patients with the syndrome are rare. These types of syndromes, which can not be solved in private dental clinic, are mostly directed to Oral, Dental, and Chin Surgery clinics. As with all syndromes, individuals with PS have specific characteristics such as turribrachycephaly, maxillary hypoplasia, ocular proptosis, hypertelorism, strabismus, exophthalmos. For this reason, the well-known knowledge of patients with syndrome by dentists is great importance to be direct of his/her family.
Figure 8. The appearance of the mandible after teeth extraction.

References