

## Pfeiffer Syndrome Type 1: A Case Report

Case Report

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## Abstract

Pfeiffer syndrome is a rare syndrome which is defined by Pfeiffer in 1964 is autozomal dominant and characterized with variable severity of brachycephalic skull, regressed midface, expansion in hands and feet, broad thumb and big toes syndactyly. This syndrome is defined as a rare syndrome with autosomal, dominantly inherited. Genetic mutations are rare but possible. The syndrome plays a role of craniosynostosis of coronal, lambdoid and occasionally sagittal suturas. Eyes are seen puffy due to shallow orbital structure and resulted by maxillary hipoplasia (ocular proptosis). Hipertelorism and down sloping palpebral fissures are common and strabismus is often seen. Intelligence is normal. In this case report, we aim to present the performed primary teeth extractions and following up in 4 years old girl with type 1 Pfeiffer Syndrome.

**Keywords:** Pfeiffer Syndrome; Syndactyly; Craniosynostosis; Congenital Anomalies.

## 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 [1]. PS is characterized by the appearance of skull and leg anomalies in eight individuals from three quadrants [2].

PS, 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 [3]. Genetic mutations are rarely possible. Patients have premature fusion of the coronal and lambdoid sutures and occasionally of the sagittal sutures, leading to an abnormal skull shape [2].

Characteristically, clinical images include; turribrakisefali (sharp rounded head shape, pronounced and protruding forehead) and maxillary hypoplasia, exophthalmos, small zygoma and undeveloped maxilla. Sometimes partial soft tissue as syndactyly also could be seen in the hand [2]. Eyes seem swollen due to shallow orbitas that is caused by maxiller hypoplasia (ocular propitosis). Hypertelorism and downward sloping palpebral fissures are common and strabismus is seen. Intelligence is normal. The broad

hand and toes are characteristic for the syndrome. It is reported with a review of literature the first neonate in Korea with PS who had bicoronal craniosynostosis, bilateral syndactyly of fingers and toes [1].

Other characteristics of PS are; lower ear lobes and finger anomalies. Intra oral features include mandibular prognathism, malocclusion, cleft pseudo-palate and bifid uvula, enlargement of the alveolar bone, high palatal arch, and crowded teeth [4, 5]. 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]. 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

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who applied to our clinic due to dental problems, turribrachycephaly, maxillary hypoplasia, ocular propitosis, hypertelorism, strabismus, exophthalmos were detected (Figure 1). According to patient record, 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 normal pregnancy period and her mother and father have first 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 (Figures 2, 3). The intraoral clinical and radiological examination revealed that oral hygiene was not good and had quite a lot caries and loss of teeth (Figures 4,5 and 6). Primary teeth, which have wide caries and 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 [2]. 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 [1]. 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 receptor (FGFR). It is known that FGFR 2 and FGFR 1 receptors are caused by mutations in exons IIIa and IIIc [7]. 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 [8]. 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 [9]. Her parents have first degree relative marriage. The syndrome is caused due to that reason according to our estimation. Bannink et al., reported that obstructive sleep apnea developed in approximately 50% of patients with Apert, Crouzon, or PS due to moderate hypoplasia [10]. Boutros et al., reported in patients with Apert, Crouzon, and PS, a narrowed cranial tendon caused narrowed condylar area, and mandibular anomalies appeared in the form of decreasing bicondylar width and bigonial width is normal [11]. In our case, only turribrachycephaly, maxillary hypoplasia, ocular proptosis, hyper-



**Figure 1.** 4-year-old girl turribrachycephaly, maxillary hypoplasia, ocular proptosis, hypertelorism, strabismus, exophthalmos.



**Figure 2.** Fingers.



**Figure 3.** Toes.



Figure 4. Caries on primary teeth in maxilla.



Figure 5. Caries on primary teeth in the mandible.



Figure 6. Orthopantomographic image of the case.



Figure 7. The appearance of the maxilla after teeth extraction.

telorism, strabismus, exophthalmos have been observed and still following to detect possible anomalies. Patients that referred in the dentist clinic are often healthy individuals; patients with the syndrome are rare. These types of syndromes, which could not be solved in private dental clinic, are mostly directed to Oral and Maxillofacial Surgery clinics. As with all syndromes, individuals with PS have specific characteristics such as turribrakisephal, maxillary hypoplasia, ocular proptosis, hypertelorism, strabismus, exophthalmos. For this reason, to direct his/her family correctly,

having respectable knowledge about the PS by dentists is a must.

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**Figure 8. The appearance of the mandible after teeth extraction.**

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