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Case Report

An unusual case of tricho–dento–osseous syndrome

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ABSTRACT

Tricho–dento–osseous (TDO) syndrome is a multisystem congenital disorder that is known by bone, skin, and hair abnormalities. Primitive studies show different varieties of manifestations related to this disorder, which involve sclerotic bones, nail involvement, enamel hypoplasia, mandibular prognathism, and taurodontism. Although exploring different TDO cases revealed genetic mutations in all of them, they have many variations in phenotypic view. In this study, we report a case whose primary diagnosis was alopecia and came for extraction of her third molars, but after clinical and radiographic examination, it was found that the cause of her disease was something different.

Key Words: Alopecia, genetic diseases, tricho–dento–osseous syndrome

INTRODUCTION

Tricho–dento–osseous syndrome is identified as a rare congenital syndrome, which manifests as autosomal dominant and is belonged to the group of diseases, named as ectodermal dysplasia.[1,2] This disorder is mainly caused by DLX3 gene mutation on the 17q21 chromosome, which is associated with bone, hair, and nail development.[3] From phenotypic viewpoint, tricho–dento–osseous (TDO) clinical signs indicate many varieties, therefore may be missed and true diagnosis of this disorder is difficult. However, this hypothesizes that the clinical signs variety is because of genetic heterogeneity or phenotypical variety has not been proved yet.[1,4] The most common clinical findings of TDO are thin hairs (curly at birth and stiffening with age increasing), enamel hypoplasia, taurodontism (especially molars), and increase in bone density (especially skull).[3,5]

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CASE REPORT

A 21-year-old female who visited her dentist, referred to department of surgery, School of Dentistry, Isfahan University of Medical Sciences, to extract her semi-impacted third molars. Investigating her medical background did not indicate any signs of systemic disease. The patient suffered only from severe hair losing, and the dermatologist's diagnosis was alopecia. Investigating familial history indicated no special sign. Clinical examination showed the head hair loss, but the body and face hairs were not lost. Further studies revealed that the patient had normal hair at childhood, but she lost them gradually [Figure 1].

The nail and skin were normal, and it was observed no kind of abnormality [Figure 2]. In the examination of the neck, the lymph nodes were normal, and

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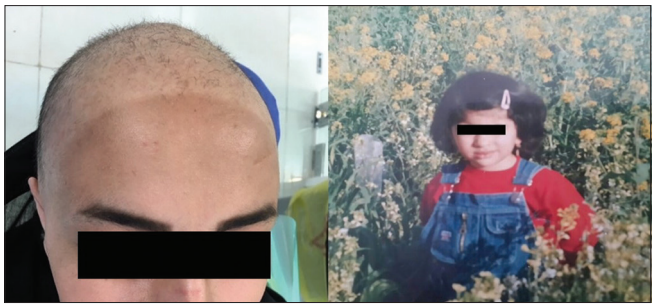

Figure 1: Normal hair in childhood and hair loss in adulthood.

Figure 2: Normal nail and skin.

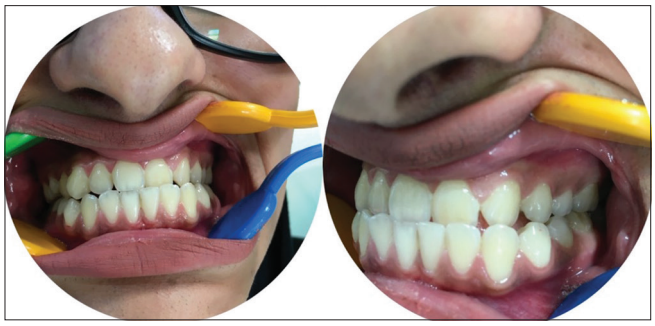

Figure 3: Prognathism of mandible and Class III relationship.

mandibular prognathism was observed in facial examination [Figure 3].

Oral mucus and tonsils were normal and there was no inflammation. The teeth of the patient did not have any special anomaly. The teeth color and shape were in a normal condition. Bilateral mandibular third molars were semi-impacted, and the first mandibular molar restoration and the second mandibular molar occlusal caries were observed [Figure 4]. In panoramic view, the roots of all teeth were very short [Figure 5]. Therefore, the TDO syndrome was considered, and the patient was referred to genetic investigation, due to available abnormalities in hair, teeth, and bone. Genetic analysis indicated a mutation in the DLX3 gene. Genetic linkage was observed on chromosome 17q21 and a 4 base-pair deletion has been identified in the DLX3 gene.

DISCUSSION

Alopecia is the most frequent autoimmune disease with unknown etiology and pathogenesis, and rarely has the teeth origin.[6,7] The autoimmune diseases such as vitiligo, thyroid disease, and atopic are along with alopecia; however, genetically bases exist for them.[7] A theory said the infection originated from fungus, bacteria, and virus or their toxins, which are originated from infection centers like oral cavity, tonsils, adenoid, and sinus, can be considered as the causes of this disease. Although there is no certain agreement about alopecia etiology and pathogenesis, immunologic base is the most popular hypothesis.[6] Teeth infection can rarely a source for alopecia.[6] Whereas, in this case systemic examination, no kind of infectious or autoimmune disease were observed.

TDO is a rare autosomal dominant systemic genetic disease, which is created as a result of heterozygous mutations in DLX3 and also has an unknown incidence rate.[8]

The disease diagnosis criteria are: generalized defects of enamel, sever taurodontism (especially in the mandible first permanent molars), autosomal dominant instinct, and one of the minor manifestations. These manifestations include nail defect, sclerotic bone, and wavy, kinky and curly hairs at childhood, which can be straighten out in the future.[1] Panoramic radiography needs to be prescribed routinely for the cases in which clinical signs of jaw dysfunction are observed.[9,10] However, these signs and symptoms are frequent at different clinical manifestations.[3]

There are significant variety to describe this syndrome in pervious literature.[11,12] The investigations of different families with TDO, demonstrated that there are various differences for TDO appearances even between members of a family.[13,14] As a result, with respect to phenotypic variety, many subtypes were proposed for TDO (TDO I, TDO II, and TDO III).[13] The classical type of TDO or the mild version of the disease with different phenotypes can accompany DLX3 mutation. Amelogenesis imperfecta and taurodontism appear to be the general features of all the cases, and they have been reported. The skin, hair, bone, and nails are also reported.[8]

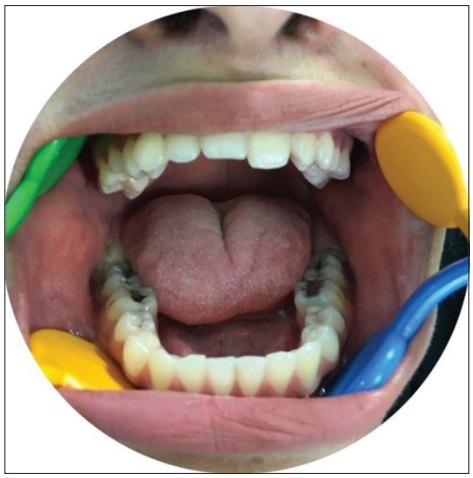

Figure 4: Intraoral photograph. The crowns of teeth are normal.

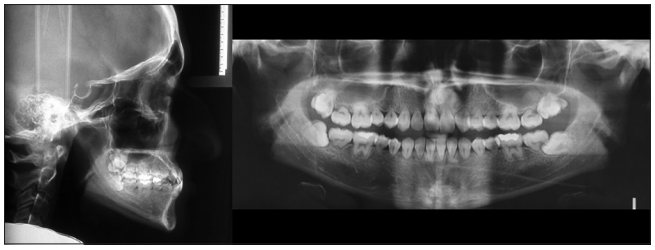

Figure 5: Panoramic and lateral cephalometric view. Severe shortening roots of all teeth and mandibular prognathism are observed.

Although studies expressed that soft, hypoplastic, or pitted enamel is a hallmark for TDO, they acknowledged that the genetic heterogeneity and environmental factors, that caused TDO phenotypic variety, must not be ignored.[11] In this case, no enamel involvement was observed.

Another cephalometric study about the cranial parameter varieties of a 40-member family (struggling with TDO) demonstrated that increasing the mandibular body length might be the only bone parameter for TDO.[15] In fact, mutation of DLX3 gen in TDO syndrome is accompanied with significant increasing of the mandibular body length.[4] According to the terminal difference of each kind of cell, DLX3 indicates different effects on mineralized tissue. In addition, direct involvement of DLX3 was observed in the osteoprogenitor cells that express the bone matrix protein (e.g., collagen type 1), bone sialoprotein, osteocalcin, and alkaline phosphatase.[16] The bone thickening in TDO can increase either fracture risk or macrocephaly.[1]

Wright *et al*. in another study conducted on 33 patients reported that kinky/curly hairs were existed in 85% of patients at birth time. In 54% of patients, the kinky hairs disappear after the infancy period, but they remain as coarse thick in other patients.[17] Price *et al*. reported that the phonotype variety in Virginia and North Carolina revealed that clinical variation is not caused by genetic heterogeneity at the major locus, but can be considered as a result of genetic heterogeneity at other epigenetic loci and/or the environment factor collaboration.[12] Moreover, Mayer *et al*. reported an atopic dermatitis in a TDO case.[3]

Moghim Farooji *et al*. reported a 10-year-old case of TDO and consumer of immunosuppressive. Due to the late referring, some of the teeth had pulp lesion. They declared that regular follow-up and on time dental treatment especially in critical age, prevents pulp lesion in patients with TDO syndrome.[18]

Harbuz *et al*. reported a familial case of TDO with osteogenesis imperfecta and intellectual disability, which had heterozygous deletion of DLX3.[14] For dentists, the TDO importance is because of two reasons: severe hypoplastic defects of enamel and difficulty in the diagnosis of hypomature and hypoplastic amelogenesis imperfecta with taurodontism of this syndrome.[1] Among teeth abnormalities, thin enamel (0.125–0.25 of normal thickness) is observable in these patients, which are mostly appear in primary teeth and fewer in permanent teeth.[1,2]

TDO syndrome management is team approach. Because the most frequent problem of this syndrome is associated with the teeth and longtime follow-up of these patients is quite needed, dental team plays an important role in this approach.[1]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

The authors of this manuscript declare that they have no conflicts of interest, real or perceived, financial or nonfinancial in this article.

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