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Case Report

Witkop's syndrome: A case report

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ABSTRACT

Witkop's syndrome is a rare autosomal dominant disorder characterized by tooth and nail abnormalities. It is a variant of ectodermal dysplasia, which is thought to occur in approximately 1 in 100,000 live births. Thus, such a low occurrence and paucity of clearly delineating features make its identification in general population a real challenge. This case report aims to add to the existing literature a commonsense approach of identifying the uncommon thing.

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1. Introduction

Agenesis of 6 or more teeth, excluding third molars is called oligodontia. Its prevalence has been reported as 0.3%,¹ with female-to-male gender ratio of 3:2.² Oligodontia could be isolated or be part of syndromes such as ectodermal dysplasia (ED), Rieger's syndrome, oto-palato-digital syndrome, Witkop syndrome, oro-facial-digital syndrome, oculo-facial-cardio-dental syndrome, or incontinentia pigmenti.³

ED is the term used to describe a group of rare inherited disorders characterized by dysplasia of tissues of ectodermal

origin with a reported prevalence of approximately 1 in 100,000 live births. Out of the 117 varieties of ED as described by Freire Maia and Pinheiro, one is the 'tooth and nail syndrome' (TNS), which was first described by Witkop in 1965. It is a rare autosomal dominant disorder characterized by hypodontia and morphological changes in teeth, along with dysgenesis of nails.^{4–7}

The congenital absence of teeth can seriously disable the young patient physically and emotionally, especially during adolescence. Retention of primary teeth in the absence of permanent successors is of significance, since they help retain the height of the alveolar bone in an area, which may be a site

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for further comprehensive therapy. The aim of this article is to report a case of Witkop's syndrome and add to the existing literature, as it has quite a low rate of occurrence and paucity of clearly delineating features making its identification in general population a real challenge.

2. Case report

A 23-year-old male patient visited the Department of Prosthodontics, Crowns and Bridges for seeking treatment for his malformed teeth. He remembered having small teeth since the time they erupted.

Medical history was negative for extremely high fevers, and he was normothermic. The patient did not report ophthalmologic lesions including cataracts, microphthalmia, optic atrophy, strabismus, retrolental fibroplasia, central nervous system involvement, atrial septal defect and/or ventricular septal defect, or mitral valve prolapse, and lesions of the skeletal system. There did not occur any erythematous and vesiculobulbous lesions on the trunk and extremities. The patient did not have hypospadias and anal stenosis. Biochemical analysis was normal. No eosinophilia was reported.

General examination revealed marked longitudinal ridging of the nail plates of all 10 fingers was noticed. The toenails were more affected with koilonychia, and had thin nail plates (Fig. 1). He reported presence of similar findings in 2 of 3 siblings, 1 elder brother and 1 sister from her maternal aunt.

Oral examination revealed presence of 13, 22, 71, 72, 73, 35, 36, 81, 83, 45, and 46 (Fig. 2) and root stumps of 54, 55, 56, 64, 65, and 66. The alveolar process on edentulous ridge posterior to 36 and 46 was undeveloped. The deciduous dentition was hypoplastic with conical teeth. Clinically, the mandibular teeth showed attrition, and tenderness on percussion was

Fig. 1 – Picture of hand and foot nails.

Fig. 2 – Intraoral view with orthopantomograph.

observed in 64 and 65. OPG examination revealed periapical radiolucencies around 64, 65, 71, 81, and 83. He had not taken any treatment for his present oral condition.

Considering the clinical findings, he was diagnosed to be a patient of Witkop's syndrome (familial hypodontia, fingernail onychorrhexis, and toenail koilonychia).

3. Discussion

A number of factors can lead to congenital oligodontia: intrauterine problems, space limitation, physical obstruction or disruption of the dental lamina, failure of initiation of the underlying mesenchyme. In humans, mutations in genes *MSX-1*, *PAX9*, and *PITX2* also result in missing teeth according to specific patterns. Studies have shown that *MSX1* and *PAX* genes play a role in early tooth development. *PAX* plays a role in the absence of wisdom teeth in some human populations, and mutations in *MSX* have been associated with non-syndromic cleft lip with/without cleft palate and autosomal dominant hypodontia. *MSX1*-associated oligodontia typically includes missing maxillary and mandibular second premolars and maxillary first premolar. A distinguishing feature of *MSX*-associated oligodontia is the frequent absence of maxillary first premolars, while *PAX*-associated oligodontia is associated

with an absence of the maxillary and mandibular second molars. Dental anomalies related to hard tissue formation appear to be related to genes coding for structural proteins contributing to the formation of enamel and dentin matrices or to the proteinases capable of degrading these matrices.^{3–5} Children with conical teeth, hypodontia or anodontia without some other readily apparent etiology should undergo a detailed history and examination to determine the differential diagnosis. Obtaining a family history is important, as Fried's tooth-and-nail syndrome looks very similar to Witkop's tooth-and-nail syndrome clinically but is inherited in an autosomal recessive fashion.

In the present case, the patient was normothermic with normal sweat gland function. Therefore, the differential diagnosis of ED was ruled out. Negative findings on examinations of cardiovascular, central nervous, skeletal systems and on skin ruled out the possibilities of Rieger's syndrome, otopalato-digital syndrome, oro-facial-digital syndrome, oculo-facial-cardio-dental syndrome, or incontinentia pigmenti.

Witkop syndrome is a variant of ED. Mutations in *MSX-1* cause failure and development of dentition in this syndrome.⁶ There is hypodontia and, infrequently, anodontia of the permanent teeth; seldom more than 20 permanent teeth are missing. Absent maxillary incisors, second molars, and maxillary canines are the most common missing teeth. Defects in primary teeth may be so mild that no abnormality is noted until the permanent teeth fail to erupt. Typically, dental anomalies are less severe in the tooth-and-nail syndrome, when compared to the more common x-linked recessive hypohidrotic EDs.⁶

The tooth-and-nail syndrome is also manifested by defects in the nail plates of fingers and toes. The nail defects are, however, alleviated with age and may not be easily detectable during adulthood. Typically, toenails are more severely affected than fingernails, and in some instances, nail plates are absent at birth.⁴

Hair defects are not specifically associated with Witkop's syndrome, but some patients might have finely textured or diffusely thin hair but neither marked hypotrichosis nor areas of alopecia are seen.⁷

Anomalies of facial topography, such as frontal bossing, depressed nasal ridges, and everted lips, commonly seen in patients with x-linked recessive ED, are not seen in Witkop's syndrome. Sweat gland function is normal. In general, the features of the tooth-and-nail syndrome are much less severe than those in more common EDs, so that it may be easily missed by an unsuspecting clinician.⁷

Usually with genodermatoses, no specific treatment is available. Other EDs like Curry-Hall syndrome and deafness and onychosteodystrophy with retardation (DOOR) syndrome

display defects confined to the nails and teeth, although these children are characteristically more severely affected. Toddlers and young children manifesting any of these diseases should be evaluated by a neurologist for retardation and seizures (Curry-Hall) and an audiologist. Genetic counseling should be mandatory for both the parents, if either one has a family history of genetic disorder, and affected children should be under regular evaluation by a pediatrician.⁷

4. Conclusion

Identification of Witkop's syndrome patient is a real challenge as with any form of ED, where young patients display a range of symptoms. Partial or total anodontia leads to functional defects, such as decreased chewing capacity, speech disorder and esthetic problem. As such multidisciplinary approach to oral rehabilitation in the early stages of life may provide functional and esthetic restoration as well as psychologic health.

Conflicts of interest

The authors have none to declare.

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