Case Report

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WITKOP'S SYNDROME: A RARE ENTITY DISCUSSED

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ABSTRACT:

Witkop's syndrome also known as 'tooth & nail syndrome' or 'nail dysgenesis & hyodontia' is a rare autosomal dominant disorder characteristically presenting hypodontia& morphological changes in teeth alongwith dysgenesis of nails. The incidence of Witkop's syndrome is 1-2 in every 10,000 individuals. The present case describes a 17-year old male patient showing characteristic features of Witkop's syndrome and multifaceted treatment provided to the patient.

Keywords: Witkop's syndrome, MSX1 gene, hypodontia, nail dysgenesis.

INTRODUCTION

Witkop's syndrome is a rare autosomal dominant disorder which was first described by Witkop in 1965.[1] The mesenchyme gene MSX1 which is a transcription factor expressed in several embryonic structures, was proved to be responsible for etiology of Witkop's syndrome in 2001.[2-4] It is characterized by hypodontia and morphological changes in teeth, along with dysgenesis of nails.[5-8] It is also known as the 'tooth and nail syndrome' (TNS) or 'nail dysgenesis and hypodontia'.[9] The clinical feature associated with Witkop's syndrome are hypodontia and infrequently anodontia of the permanent teeth in which seldom more than 20 permanent teeth are missing.[10]

CASE DETAIL:

A 17-year-old male patient reported to our institution with the chief complaint of small teeth, generalized spacing between teeth. Patient gave no positive history of exfoliation or extraction of teeth but gave a history of delayed eruption of teeth. When asked about his family history he gave a positive history of his father suffering from a similar complaint. He also gave a history of slow growth of his toe nails, which also frequently tended to fracture.

A general examination revealed normal hair (both in growth and texture), eyebrows and skin. **(Figure1).** He had dysplastic toe nails, no abnormalities were detected in the finger nails **(Figure 2).**

On intraoral examination, 20 retained deciduous and 1 permanent teeth (left maxillary first permanent molar) were present. All other permanent teeth were

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missing clinically. All teeth were attritedand generalized spacing was present in both arches. Loss of vertical dimension was noted. High frenal attachment was seen in upper anterior region. **(Figure 3).**

OPG **(Figure 4)** revealed resorption of the roots in both upper and lower anterior region.Sobased on the above extraoral& intraoral clinical findings, positive familial history, a provisional diagnosis of Witkop's syndrome was made.

TREATMENT: After keeping in mind all the needs and limitations of the adolescent patient & in order to provide esthetics and optimum function, a multifaceted approach was planned with cooperation of Periodontist, Prosthodontist&Endodontist.Patient was advised conservative tooth supported full mouth rehabilitation, endodontic treatment including RCT for 51,52,53,61,62,63 along with post and core for 53,63, frenectomy in upper anterior region followed by crown lengthening from 53 to 63 region **(Figure 5 & 6).**

DISCUSSION

Witkop's syndrome is rare autosomal dominant disorder and variants of ectodermal dysplasia which occur approximately one to two in every 10,000 individuals.[11] In Witkop's syndrometooth agenesis is accompanied by nail deformities such as the nail plates are defective and thinner.[2] In most individuals with Witkop's syndrome toenails are severely affected than finger nails and in some cases nail plates are absent at birth.[5] Nail dysplasia is a common feature in the Witkop's syndrome where nails tend to be spoon – shaped (koilenchyia), thin, slow growing, brittle & easy to break (onychorrhexis). The nail defects are however alleviated with age and may not be easily detectable during adulthood.[7, 9, 12-14]

It is characterized by hypodontia and infrequently anodontia of the permanent teeth. Maxillary incisors, canines and second molars are most commonly missing and the affected teeth tend to have narrow crowns & conical shape are widely spaced.[15-16] In Witkop's syndrome partial or total agenesis of dentition is sometime present causes over retention of the primary teeth.[17]

Various genetic syndrome in which oligodontia occurs include Ectrodactyly-Ectodermal Dysplasia-Clefting syndrome, Cleft Lip Palate Ectodermal Dysplasia syndrome, Oral Facial Digital syndrome type I , Witkop Tooth-Nail syndrome, Fried syndrome, Hair- Nail- Skin- Teeth dysplasias, Ectodermal Dysplasia, IncontinentiaPigmenti, Down syndrome, Rieger syndrome, Wolf-Hirschhorn syndrome, Van der Woude syndrome.[18-19]

Patient suffering from oligodontia are mainly affected with significant psychological, functional and esthetic problems.[20] In the present case, the patient was normothermic with normal sweat gland function. As compared to x-linked recessive hypohydrotic ED, the dental anomalies are less sever in Witkop's syndrome.[7] Hair defects like

hypotrichosis & alopecia are not specifically associated with Witkop's syndrome & 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.[8]

Dental treatment can vary depending on the severity of the disease and generally requires a multidisciplinary approach.

In our case, factors to be taken in consideration before planning treatment are age of the patient, number & condition of retained teeth, condition of supporting tissue, occlusion & interocclusal space.

Non surgical treatment plan was adopted as it was cost effective as compared to implant prosthesis.

The patient was satisfied about his improved appearance & masticatory function **(Figure 7).** Presently he is on periodic regular follow up.

CONCLUSION

Witkop's syndrome is a rare disorder where patients present with hypodontia leading to functional disability. So, we as dentists are accumstomed to provide a proper diagnosis & multifaceted treatment to restore the functional defects & provide a psychological boost by restoring proper esthetics to the patient.

Conflicts of interest: No conflicts of interest recorded

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FIGURES:

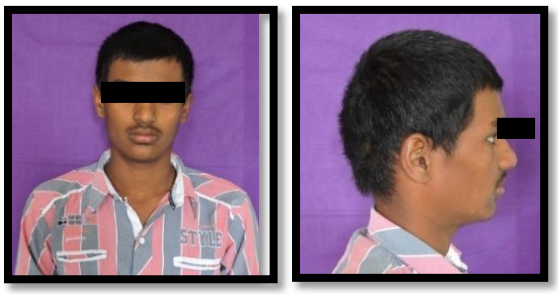

Figure 1 - Front & lateral view of patient shows normal scalp hair.

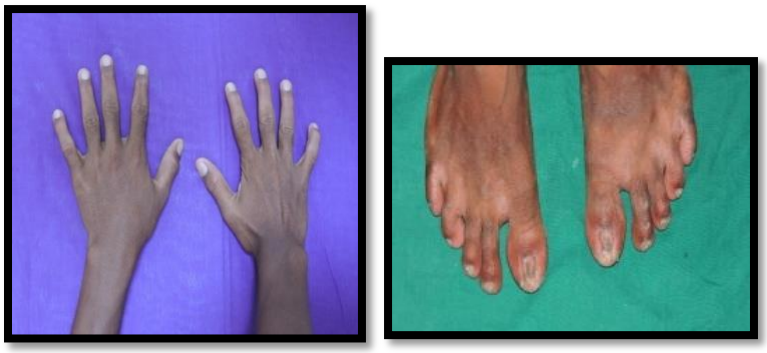

Figure 2 – A) Normal finger nails

B) Dysplastic toe nails.

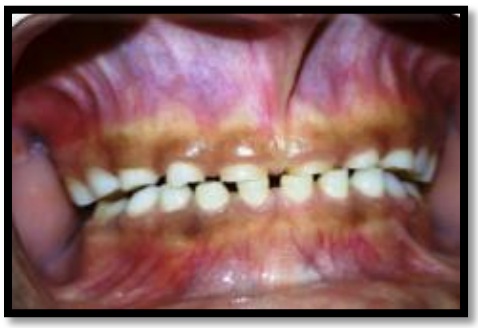

Figure 3 – preoperative intraoral photograph.

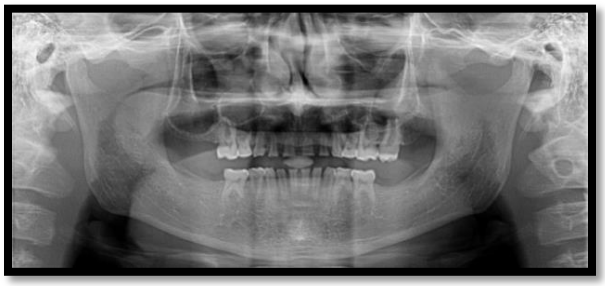

Figure 4 – OPG showing congenital absence of tooth buds except 26.

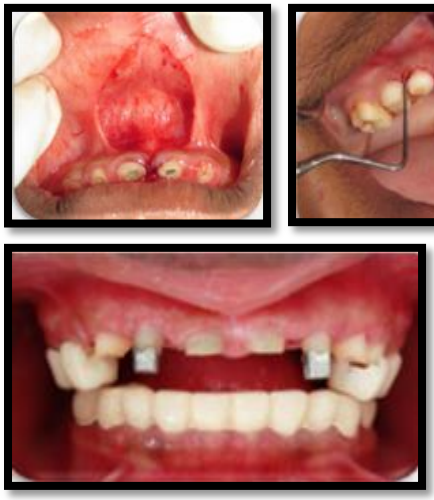

Figure 5 – A) Frenectomy B) Crown lengthening C) Post preparation.

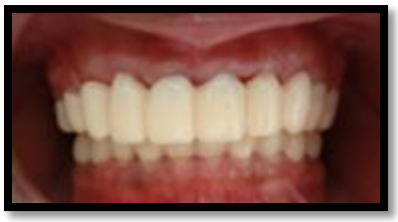

Figure 6 – Photograph of intraoral view showing FPD in occlusion.

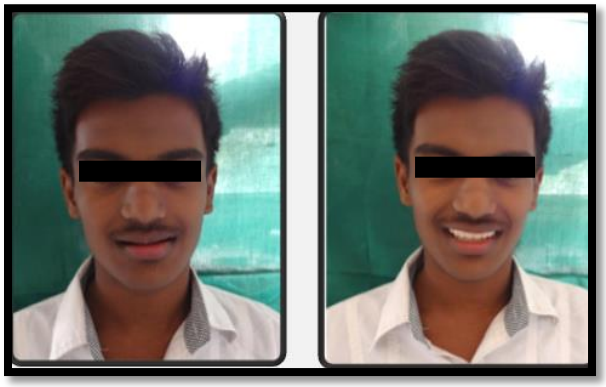

Figure 7 – Preoperative & postoperative photographs of the patient.