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Case Report

Waardenburg Syndrome Type 2: A Restorative Perspective

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Abstract

Waardenburg syndrome Type 2 has not been documented in the Asia-pacific region, here we present a case with no apparent family history.

Keywords: *Waardenburg syndrome (WS); Sensorineural hearing loss; Autosomal Dominant*

Introduction

Inherited hearing loss is most commonly found in the Waardenburg syndrome (WS). Various degrees, depigmentary abnormalities of the hair, eyes and skin are associated with this syndrome [1]. This syndrome is determined by the absence of melanocytes from the eyes, hair and skin [2]. Waardenburg syndrome was first described in 1951 by Petrus Johannes. Waardenburg II has an estimated prevalence of one in every 42,000 births [1,2]. The genders are equally affected. Six genes are involved in this syndrome: PAX3 (encoding the paired box 3 transcription factor), MITF (microphthalmia associated transcription factor), EDN3 (endothelin 3), EDNRB (endothelin receptor type B), SOX10 (encoding the Sry BOX10 transcription factor) and SNAI2 (snailhomolog 2). These six genes are affected with variable frequency in WS. Waardenburg syndrome appears to be associated with the defective migration of neural crest cells during the third month of pregnancy. These neural crest cells eventually give rise to melanocytes, sympathetic ganglia, sensory components of the cranial and spinal nerves, and membranous bones of the face and the palate, dentin and visceral ganglia [3,4]. This case report describes an individual diagnosed with WS and the restorative implications and other esthetic considerations.

Case Report

A 14 year old boy patient reported to the dental OPD of the department of conservative dentistry and endodontic's with the chief complaint of fractured upper anterior teeth and wanted restoration.

The boy had a white for lock and bright blue eyes. A written informed consent was obtained from the patient's parents, including lawful use of any pictures or images of the boy. All principles and ethical guidelines for medical research on human subjects were considered during this study.

Clinical examination

Clinical evaluation was performed , under hygienic conditions (medical scrub suits, caps, latex gloves and masks), in the presence of ambient light, using a dental mirror, cheeks retractor and a dental probe, which had been previously sterilized in autoclave conditions at 135°C.

Extra oral examination

On the frontal left side, a white forelock of hypopigmented hair was noted. The characteristic disturbance of iris pigmentation with brilliant blue irides was observed in the patient (Figure 1). He has synophrys-confluent eyebrows, hypoplastic alae nasi, narrow nostrils and broad nasal root. From the frontal aspect, the patient has a narrow facial appearance, with a slight mandibular asymmetry to the left. He has a flattened profile with a short upper lip, a thick lower lip and lack of philtrum.

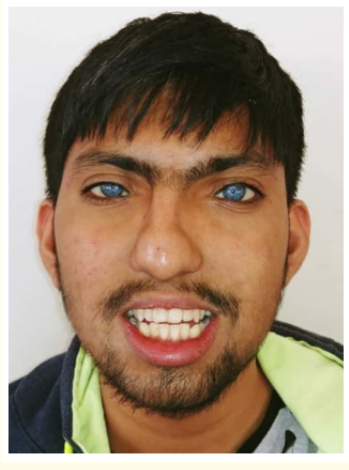

Figure 1: *Frontal aspect of patient with brilliant blue irides, syndophurs, broad nasal root and narrow nostrils*

Intra oral examination

The patient showed a Class II division 1 malocclusion, with proclined upper incisors and increased overjet, with class I canine relationships on both sides. Upper and lower midlines were not coincident. The facial appearance was reflected in the maxillary arch form, which was assessed as narrow and ovoid with a narrow maxilla and high arched palate.

Treatment

After analyzing the patient and discussing with the family the restorative options was explained and written consent obtained. As 11 was endodontically treated in the pediatric department of our hospital with a satisfactory obturation a composite corno-radicular restoration was done which was esthetically pleasing to the patient with the further option of ceramic crown if required. A few class I cavities were restored with glass ionomer Type IX owing to the poor oral hygiene and recalled once in 6 months for further evaluation. The follow up appointments after 60, 120 and 180 days have revealed an improved oral and general status of the patient and thus a better quality of life.

Figure 2: *Side aspect of patient with flat profile, short upper lip, thick lower lip.*

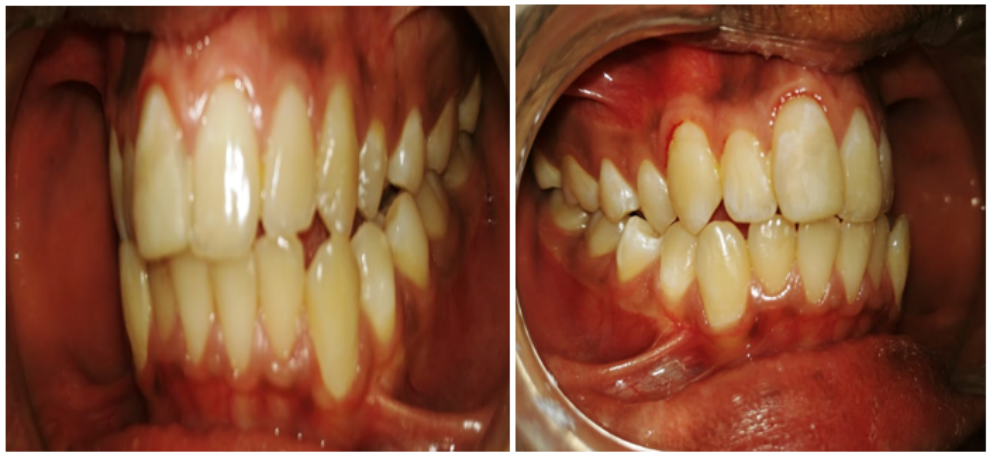

Figure 3 and 4: *Intraoral left and right with canine guided occlusion.*

Discussion

WS is associated with deafness and other abnormalities due to defective migration of neural crest derived tissues. Abnormalities in this process can explain all of the clinical signs of WS that have been described in literature. Individuals with WS may always not present with the full spectrum of clinical symptomology [5,6] associated with WS. According to the diagnostics criteria proposed by the diagnostic consortium of a person diagnosed with Wardenburg Syndrome must present with two major criteria or one major and two minor criteria. Our case has three major criteria sensorineural hearing loss (SNHL), White for lock, abnormal pigmentation of iris (brilliantly blue).

In our case report the patient suffers from congenital deafness, which may be polyetiological: WS, Usher's syndrome or the mutation of connexin 26 gene, SNHL can be explained by lack of melanocyte-derived intermediate cells of stria vascularis found in the cochlea. This absence induces endolymphatic collapse, culminating with atropy of corti's organ, a process that is called cochleamacular degeneration [7,8]. The most obvious of WS is the white forelock which is seen in our patient too since birth. In WS patients the facial aspect is quite characteristic, with hypoplastic nose, broad nasal root and alar cartilages, which leads to narrow nostrils, which is the cause mouth breathing in the patient. In our patient too eyebrows are confluent at the midline. Congenital deafness is the most and significant clinical symptom. Early diagnosis is key to the successful treatment of WS patients. In our case report we have done only restorative treatment and counseled the family regarding dental treatment and interdisciplinary medical intervention [9-12].

Conclusion

We have reported a case of Waardenburg Syndrome Type 2, a very rare disease, with mouth breathing and congenital deafness. As it is very rare to report such cases, especially in the Indian population a comprehensive multidisciplinary medical and dental is required as well as periodic follow up for the successful treatment.

Conflict of Interest

The authors declare that they do not have any conflict of interests.

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