International Journal of Applied Dental Sciences 2022; 8(3): 116-117

ISSN Print: 2394-7489
ISSN Online: 2394-7497
IJADS 2022; 8(3): 116-117

www.oraljournal.com
Received: 15-05-2022
Accepted: 17-06-2022

Dr. Pulkita Prakash
PGT Student, Buddha Institute of Dental Sciences and Hospital, Patna, Bihar, India

Dr Amitabh Kumar Sinha
PGT Student, Buddha Institute of Dental Sciences and Hospital, Patna, Bihar, India

Dr. Anmol Ratan
Government Medical officer Bihar, India

Corresponding Author:
Dr. Pulkita Prakash
PGT Student, Buddha Institute of Dental Sciences and Hospital, Patna, Bihar, India

A rare genetic disorder: Waardenburg syndrome

Dr. Pulkita Prakash, Dr Amitabh Kumar Sinha and Dr. Anmol Ratan

DOI: https://doi.org/10.22271/oral.2022.v8.i3b.1594

Abstract
Waardenburg syndrome is an inherited autosomal dominant disorder consists group of rare genetic conditions. Mutation in the PAX3, MITF, SOX10 and SNAI2 genes are known as main cause for it. There are four types of Waardenburg syndrome differing in phenotypic characteristics are described. This syndrome is characterized by congenital deafness or some degree of hearing loss, different color of eyes with various degree of depigmentation of skin and hair, increased inter-canthal distance, Dystopia Canthorum. In this report a case has been discussed of finding a 30 years old male patient with Waardenburg syndrome.

Keywords: Waardenburg syndrome, autosomal dominant disorder, congenital deafness, depigmentation, heterochromia

Introduction
Waardenburg syndrome is a rare genetic disorder most often characterized by varying degree of deafness, minor defects in structures arising from neural crest, and pigmentation anomalies [1]. It was described first by Petrus Johannes Waardenburg in 1951. The characteristic clinical findings include sensorineural hearing loss, increased intercanthal distance, heterochromia iridis, pigmentary abnormalities of hair and skin along with dental findings of agenesis, cleft lip and palate and tooth malformations [2]. The incidence of WS is estimated as 1 on 42,000 among Caucasian populations, or 2-5% of patient with congenital deafness, and 0.9-2.8% among adults with hearing impairment [3].

Case report
A 30 years old male patient reported with the chief complaint of pain in lower left back tooth region. On clinical examination it was found that patient have bilateral hearing loss since birth, there was increased intercanthal distance, broad nasal root, heterochromia (different color of eyes) with depigmentation on different sites of body was seen. Patient didn't have any neurological disturbances. Intraoral examination revealed an increased overjet, narrow and high arched palate, and early exfoliation of teeth due to caries. So, on correlating the history given by patient and on clinical examination it was diagnosed as Waardenburg syndrome.
An OPG radiograph showed bilateral condylar hypoplasia with edentulous spaces w.r.t 36, 37, 46 secondary to exfoliation of tooth due to caries. Radiolucency was seen in coronal portion of enamel, dentin and pulp w.r.t 38 suggestive of dental caries.

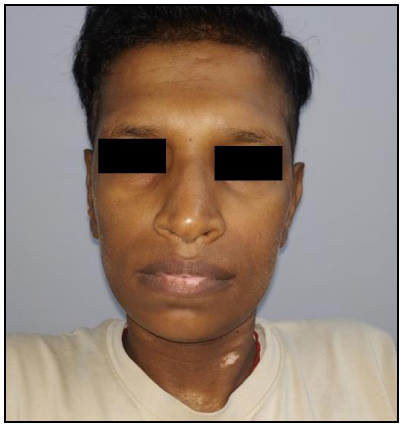

Fig 1: Extraoral profile picture of patient

Fig 2: Heterochromia

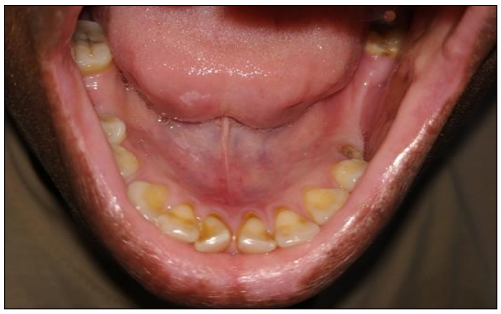

Fig 3: Intraoral picture of patient.

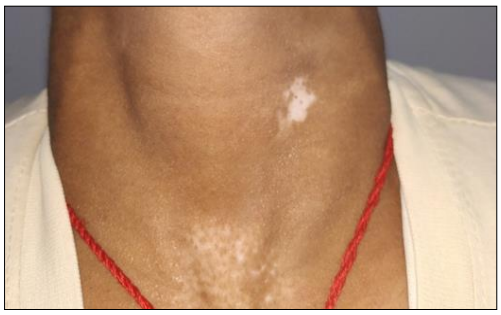

Fig 4: Showing depigmentation on neck

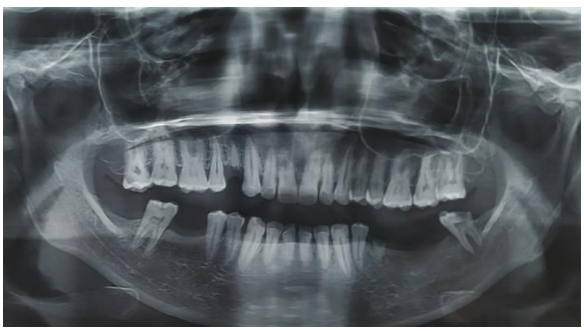

Fig 5: An OPG radiograph (red arrow) showing bilateral condylar hypoplasia.

Discussion

Waardenburg syndrome may be diagnosed at birth or early childhood or in some cases, at a later age based upon a thorough clinical evaluation, identification of characteristic physical findings, a complete patient and family history and various specialized studies [4]. Waardenburg syndrome is classified into four types depending on the additional symptoms present: WS1, WS2, WS3 and WS4. Type 1 is the classic form of WS with dystopia canthorum (lateral displacement of the inner canthi), whereas type 2 is characterized by the presence of unpigmented tissue and deafness without dystopia canthorum. Type 3 is similar to type 1 WS with additional musculoskeletal abnormalities and type 4 or Waardenburg-Hirschsprung disease is characterized by presence of an aganglionic megacolon [5].

Conclusion

Waardenburg syndrome is an autosomal dominant condition that associates with medical and dental findings. So genetic counseling for the patient will be beneficial for the patient. Early diagnosis of hearing impairment of patient with this syndrome may helpful in proper treatment.

References

1. Sudesh Kumar, Kiran Rao. Waardenburg syndrome: A rare genetic disorder, a report of two cases. Indian J Hum Genet. 2012;18:254-5.
2. Rohan Jagtap, Ambika Srivastava, Aniket Jadhav, Swati Gupta. Oral manifestation of Waardenburg syndrome: a case report and review of the literature. BJR Case Rep 2020;6:20200071.
3. Nikolay Barashkov A, Georgii Romanov P, Uigulaana Borisova P, *et al*. A rare case of Waardenburg syndrome with unilateral hearing loss caused by nonsense variant c.772C>T (p.Arg259*) in the MTF gene in Yakut patient from the Eastern Siberia (Sakha Republic, Russia). International Journal of Circumpolar Health. 78(1):1630219.
4. Luma Haj Kassem, Mohamed Fares Ahmado, Majd Sheikh Alganameh. A rare case of seven siblings with Waardenburg syndrome: a case report. J Med Case Rep. 2018;12:192.
5. Luciano Solia-Nasser, de Aquino SN, Paranaiba LMR, Gomes A, dos-Santos-Neto P, Coletta RD, *et al*. Waardenburg syndrome type 1: dental phenotypes and genetic analysis of an extended family. Med Oral. 2016;21:e321.