IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 14, Issue 11 Ver. IX (Nov. 2015), PP 04-06
www.iosrjournals.org

Waardenburg Syndrome-A Case Series

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Abstract: *A 20 months old baby referred from ENT department, as a known case of sensory neural hearing loss planned for cochlear implant for ophthalmic evaluation. On examination she had brilliant blue iris of both eyes, dystopia canthorum, hypoplastic alar nasi . With the above features, Waardenburg syndrome was suspected.*

Keywords: *Waardenburg syndrome, heterochromia iridis, brilliant blue iris, dystopia canthorum, Sensorineural hearing loss*

I. Introduction

Waardenburg syndrome (WS) is an uncommon autosomally inherited and genetically heterogeneous disorder of neural crest cell development. Incidence is 1 in 50,000 worldwide. WS does not show gender, ethnic, or racial predilection. It consists of distinctive facial and ocular features. Facial features include poliosis, broad prominent nasal root, hypertelorism. Ocular features are transilluminating irides, heterochromia iridis, confluent eyebrows, hypertrichosis, dystopia canthorum, bilateral lenticonus, mottled fundus pigmentation, increased susceptibility to dacryocystitis.[1]

II. Case Report:

Our first case was a 20 months old baby referred from ENT department for ophthalmic evaluation. She was a known case of sensory neural hearing loss planned for cochlear implant. Ophthalmic evaluation revealed a good fixation and she was able to follow the light. Ocular examination revealed dystopia canthorum, synorphrys (Hypertrophy and fusion of the eyebrows), brilliant blue iris of both eyes. She also had broad nasal root and hypoplastic alar nasi [Figure1]. Her fundus examination showed mottled hypopigmentation. She was born of full-term normal vaginal delivery. There was no history of consanguinity. Her maternal aunt (the second patient), who accompanied the baby also had similar ocular and facial features of brilliant blue iris of right eye, heterochromia iridis of left eye, synorphrys, broad nasal root and hypoplastic alar nasi [Figure2]. Her fundus examination also showed mottled hypopigmentation. Her brother [maternal aunt's son] had hypochromic iris with no other clinical features of Waardenburg syndrome which explains the varied expressibility of Waardenburg syndrome.

Our third case was a 17 year old female, known case of sensory neural hearing loss presented for routine examination in the ophthalmology OPD. She was found to have brilliant blue iris of right eye, heterochromia iridis of left eye , premature greying of hair, dystopia canthorum, broad nasal root[Figure3].

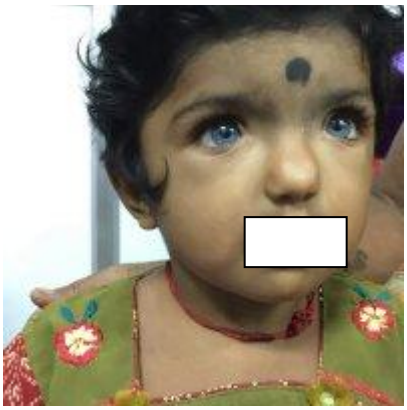

Figure1: Showing brilliant blue iris of both eyes, dystopia canthorum, broad nasal root, hypoplastic alar nasi, and synophrys - WS type1

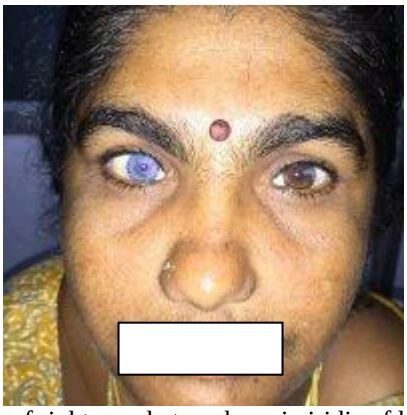

Figure2: Showing brilliant blue iris of right eye, heterochromia iridis of left eye, dystopia canthorum, broad nasal root, hypoplastic alar nasi and synophrys - WS type1

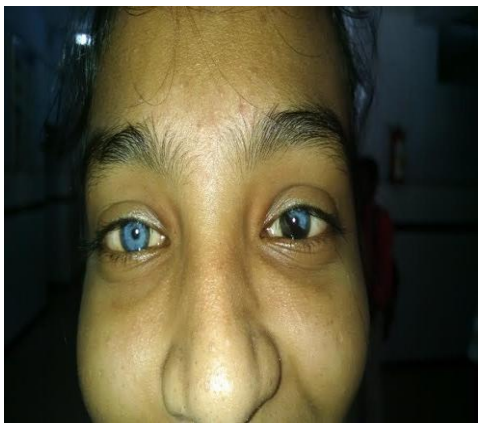

Figure3: Showing brilliant blue iris of right eye, heterochromia iridis of left eye, dystopia canthorum, broad nasal root - WS type1

III. Discussion

Waardenburg syndrome is a heterogenous disorder of neural crest cells leading to defects in the structures derived from neural crest with varying degrees of sensory neural hearing loss and pigmentation anomalies. Based on the clinical presentation, four types have been described.[2] The Diagnostic criteria for Waardenburg syndrome has been proposed by the Waardenburg consortium.[3] Accordingly two major or one major plus two minor criteria is necessary to diagnose as WS type1.

The major criteria being

1.Sensorineural hearing loss
2.Iris pigmentary abnormality (two eyes different color or iris bicolor or characteristic brilliant blue iris)
3.Hair hypopigmentation (white forelock or white hairs at other sites on the body)
4.Dystopia canthorum (lateral displacement of medial canthi and lacrimal punctum)
5.The presence of a first-degree relative previously diagnosed with WS. The minor criteria being
1.Skin hypopigmentation (congenital leukoderma/white skin patches)
2.Medial eyebrow flare (synophrys)
3.Broad nasal root
4.Hypoplasia of alae nasi
5.Premature graying of hairs (before age 30).

WS type 2 lacks dystopia canthorum.[2] WS type3 has upper limb anomalies in addition(e.g. hypoplasia, syndactyly). [5] WS Type 4 is associated with features of Hirschsprung disease in addition.[4] WS type1 and 2 are autosomal dominant, WS type 3 is sporadic or autosomal dominant, WS type4 is autosomal recessive.

Our first two cases fulfilled 4 out of 5 major criteria and 4 out of 5 minor criteria and the third case fulfilled 3 out of 5 major criteria and 2 out of 5 minor criteria. Hence a diagnosis of WS type 1 was made based on the clinical presentation and is presented for rarity.

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