Case Report

Waardenburg syndrome type-1 in a Sri Lankan patient

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Abstract

A 29-year old Sri Lankan lady, born in Manipay, Jaffna, presented with congenital deafness, brilliant blue colour iris, premature greying of hair, medial eye brow flare (synophrys) and broad nasal bridge with dystopia canthorum. We diagnosed Waardenburg syndrome (WS) type-1. Mainstay of management of this syndrome is genetic counselling and attending to sensorineural hearing loss. We arranged a hearing aid for her. It improved her quality of life and social communication.

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Introduction

Waardenburg syndrome (WS) is a rare genetic disorder characterized by congenital hearing loss, pigmentation anomalies and various ocular features including dystopia canthorum. A Dutch ophthalmologist, P.J. Waardenburg, first described Waardenburg syndrome in a patient with hearing loss, dystopia canthorum and retinal pigmentary differences in 1951 (1). There are four subtypes (type 1-4) of Waardenburg syndrome due to variable penetrance and expression of genes. Overall, the syndrome affects around 1 in 42,000 people all races and both sexes are affected equally (2). Waardenburg syndrome accounts for about 2-5% cases of congenital deafness (2). Here we report a 29-year old lady who presented with characteristic features of Waardenburg syndrome.

Case presentation

A 29-year old Sri Lankan lady, born in Manipay, Jaffna, presented with deafness and dumbness. On general examination, she had characteristic brilliant blue colour iris and premature grey hair (Figure 1). However, there were no hypopigmented patches on the skin. Medial eyebrow flare (synophrys) and broad nasal bridge with increased inter canthal distance were noticed. Intercanthal distance was 45mm, interpupillary distance was 65mm and outer canthal distance was 100mm. W index was calculated and it was 2.4 and dystopia canthorum was confirmed by these calculations. Dilated fundus evaluation showed normal fundus in both eyes without any obvious pigmentary changes. According to diagnostic criteria for WS (Table 1), she fulfilled three major and three minor criteria and we made the diagnosis of WS.

Her parents were not consanguineous. Screening of family members including parents and siblings was done. There was no ocular abnormality or deafness found.

Table 1: **Diagnostic criteria for WS as proposed by the Waardenburg Consortium**

Major Criteria
1. Congenital sensorineural hearing loss
2. Affected first degree relative
3. White forelock, hair hypopigmentation
4. Dystopia canthorum W index > 1.95
5. Pigmentary disturbances of iris:
• Complete heterochromia iridis
• Partial segmental heterochromia iridis
• Brilliant blue irides
Minor Criteria
1. Medial eyebrow flare (synophrys)
2. Congenital leukoderma: several areas of hypopigmented skin
3. Hypoplasia of alae nasi
4. Broad/high nasal root
5. Premature greying of hair

Discussion

Waardenburg syndrome is a rare non-progressive congenital genetic disorder. The diagnostic criteria for WS type-1 were proposed by the Waardenburg Consortium in 1992 (3). The individuals must have two major or one major and two minor criteria for the diagnosis of WS type-1 (Table 1). Most cases of WS type-1 are caused by mutation in the PAX3 gene located on the long arm of chromosome 2.

Based on the clinical manifestations, there are four subtypes described (4).

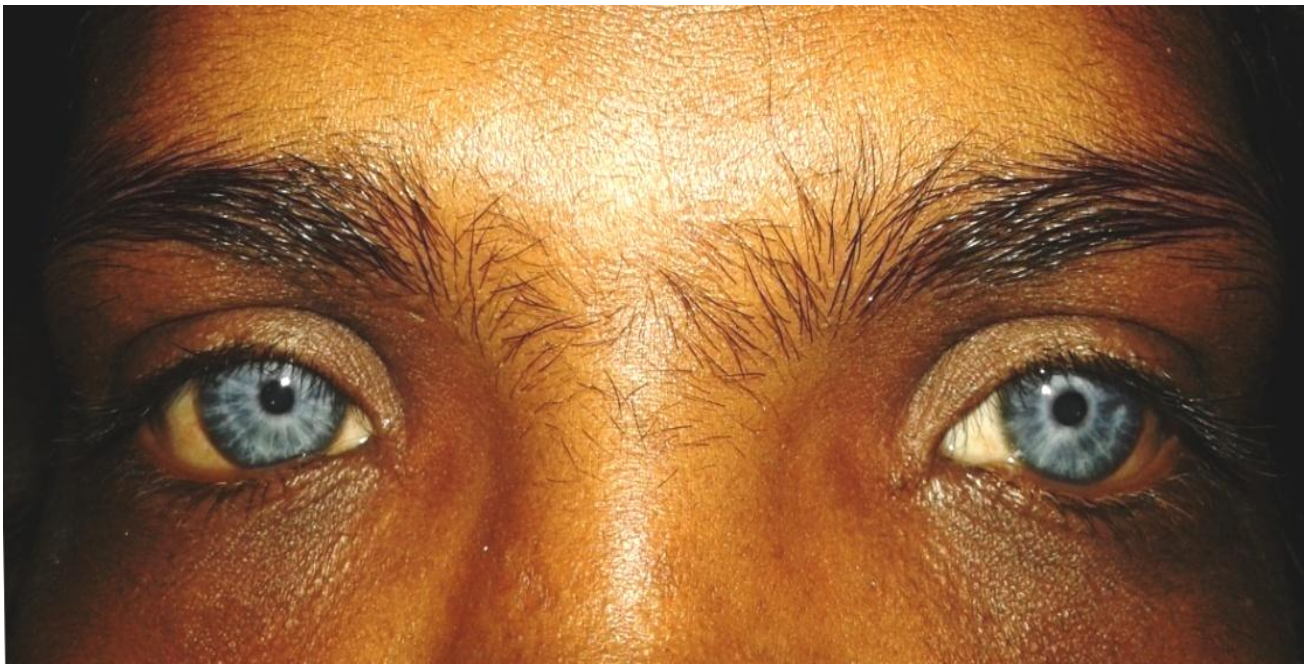

Figure 1 **Brilliant blue colour of iris**

Type-1 WS consists of dystopia canthorum and broad nasal root. Type-2 WS has all the features of type-1 but lacks dystopia canthorum. Type-3 is associated with upper limb deformities and type-4 is associated with Hirschsprung disease.

Our patient had three major criteria, namely, congenital sensorineural hearing loss, brilliant blue irides and dystopia canthorum with three minor criteria, namely, medial eyebrow flare (synophrys), broad and high nasal root and premature greying of hair. But she did not have any upper limb deformities or features of Hirschsprung disease. Based on these findings we diagnosed WS type-1. Dystopia canthorum is the most penetrant feature of WS and is found in 41.2–99% of the reported cases (5). Dystopia canthorum is defined as W index > 1.95. W index can be calculated by the following formula:

Calculate X = [2a-(0.2119c+3.909)] / c
Calculate Y = [2a-(0.2479b+3.909)] / b
W Index = X+Y+ a/b
[a = innner canthal distance, b = interpupillary distance, c = outer canthal distance]

Mainstay of management of this syndrome is genetic counselling and attending to sensorineural hearing loss by cochlear implants if detected at an early age. It will improve speech discrimination and spoken language. No special diet or activity restrictions are needed. Folic acid supplementation in pregnancy has been recommended for women at an increased risk of having a child with WS (6). For our patient we arranged a hearing aid. It improved her quality of life and social communication.

Conclusion

Early diagnosis and improvement of the hearing defects are crucial for the psychological and speech development in children with WS even though in adults, improvement of hearing by hearing aid will improve their quality of life. Tolerance and understanding of persons with WS will help support their integration into society.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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