SYNDROME OF THE STRIKING EYES: WAARDENBURG SYNDROME

Ashwini Mohandas[1], Sana Afsar[1], Gulnaz Nadri[1]

[1]Department of Pediatrics, JNMCH, AMU, Aligarh, India

Correspondence: Ashwini Mohandas, anumohan15994@gmail.com, Department of Pediatrics, Aligarh Muslim University, India.

INTRODUCTION

Waardenburg syndrome is a genetic condition inherited in an autosomal dominant fashion resulting in pigmentary defects of iris, skin and hair, congenital sensorineural deafness, and systemic abnormalities. The estimated prevalence is 1in 42,000 with the highest prevalence in Kenyan Africans and accounts for >2% of congenital deafness(1). There are four different types of Waardenburg Syndrome based on phenotypic and genotypic characteristics and is diagnosed using Waardenburg Syndrome Consortium Criteria. The Waardenburg syndrome is caused by mutations in the EDN3, EDNRB, MITF, PAX3, SNAI2, and SOX10 genes. These genes play a role in the formation and development of multiple cell types, including melanocytes. Mutations in any of these genes interfere with the normal development of melanocytes, leading to absence of melanocytes from skin, iris, hair, and stria vascularis of cochlea. PAX3 mutation is present in Type Ⅰ and Ⅲ Waardenburg syndrome. Type Ⅱ is caused by mutations in MITF and SNAI2 genes, whereas, Type Ⅳ is caused by mutations in SOX10, EDN3, or EDNRB genes(2).

CASE SUMMARY

First patient 17 months old male baby and second patient, one year old male baby, both came to pediatrics OPD with complaints of inability to speak and hear. The first baby had white forelock of hair, blue colored iris (figure 1) with hypopigmented patches, multiple hypopigmented macules on the back and chest (figure 2). The second baby had blue colored iris with no other physical findings (figure 3). Both the babies were developmentally normal assessed using Vineland Social Maturity Scale (Indian adaptation) except for the isolated speech delay. First baby had no similar family history whereas the second baby had a family history of deafness and mutism in elder brother, mother, and maternal uncle but his elder female sibling was unaffected. OAE of both the babies showed inadequate outer layer cell function and BERA was suggestive of profound bilateral hearing loss (figure 4&5). Visual acuity was 6/6 in both the babies and fundus examination was within normal limits.

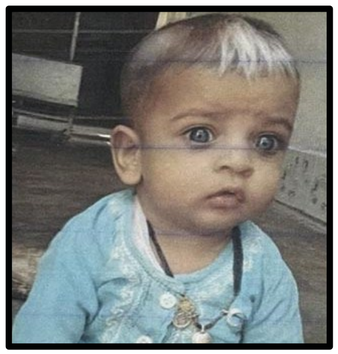

Figure : 1

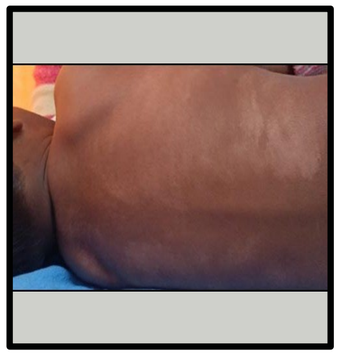

Figure : 2

Figure : 3

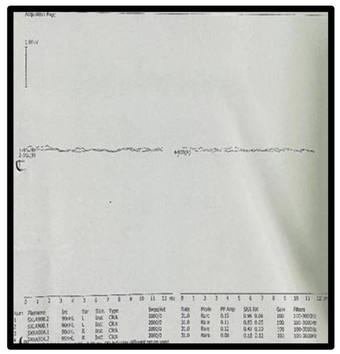

Figure : 4

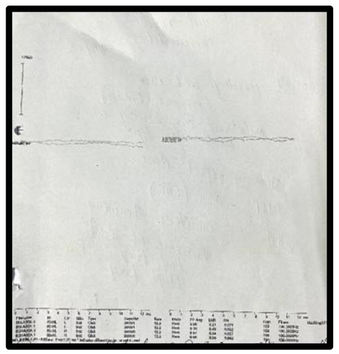

Figure : 5

DISCUSSION

Waardenburg syndrome (WS) is a rare autosomal dominant disorder characterised clinically as piebaldism, sensorineural deafness, pigmentation defects of hair, skin and iris, dystopia canthorum, synophrys, and various other defects of neural crest derived tissues (2). Not all patients of WS have all clinical manifestations. 36% to 58% of Type 1WS patients and up to 87% of Type 2 WS patients have deafness. Furthermore, hearing loss is highly variable ranging from none to severe unilateral to bilateral losses (3).

***Diagnostic criteria*:**

According to Waardenburg consortium criteria, there should be two major or one major plus two minor criteria for the diagnosis of WS (4).

MAJOR CRITERIA	MINOR CRITERIA
Congenital sensorineural hearing loss	Congenital hypopigmentation of skin
Iris pigmentation defects	Synophrys
Pigmentary defects of hair (white forelock)	Broad high nasal bridge
Dystopia canthorum	Hypoplasia of alae nasi
First degree relative with WS	Premature greying of hair

In our case series, the first baby fulfilled 3 major and one minor criteria and the second baby fulfilled 3 major criteria thus clinically fitting into the diagnostic criteria of Waardenburg syndrome.