Waardenburg Syndrome Type I: Case Report

Dr.Keya Lahiri1, Dr.Karobi Lahiri2, Dr.Rajesh Rai3, Dr.Neha Malhotra4, Dr. Pallavi Gahlowt5
M.D. Professor and Head Department Of Paediatrics, Dr. D.Y.Patil Hospital and Medical College
MS Ophthalmology, Vitero Retinal Surgeon and Paediatric Ophthalmologist, Bombay Hospital
M.D Professor, Department of Paediatrics Dr. D.Y.Patil Hospital and Medical College
M.D Resident, Department of Paediatrics, Dr. D.Y.Patil Hospital and Medical College
MD,DCH,FCPS Pediatric Intensivist, Department of Paediatrics
Dr. D.Y.Patil Hospital and Medical College

Abstract: Waardenburg Syndrome is a rare inherited oculocutaneous syndrome affecting the eyes, skin, hair and hearing. It exhibits both clinically and genetically heterogenous characteristics. We present a 2 years old male with Waardenburg Syndrome Type I, one of the four clinical types of Waardenburg Syndrome.

Keywords: oculocutaneous, congenital hearing loss, white forelock, Waardenburg Syndrome index

I. Introduction
Waardenburg Syndrome is an uncommon autosomally inherited disorder of neural crest development. It affects 1 in 42,000 individuals. It was named after a Dutch ophthalmologist, P.J Waardenburg, who identified a syndrome with six characteristic features: lateral displacement of the medial canthi, broad and high nasal root, hypertrichosis of the medial eyebrows, partial or total heterochromia iridis, white forelock and congenital deaf mutism. This condition is now categorized as Waardenburg Syndrome I. At present, four subtypes (I to IV) with variable penetrance of different clinical features have been described. Waardenburg Syndrome II lacks dystopia canthorum. Waardenburg Syndrome III is similar to Type I with additional musculoskeletal abnormalities (upper limb hypoplasia, syndactyly). Waardenburg Syndrome IV additionally has Hirschsprung’s disease.

Case Report:
A 2 year old male, born of third degree consanguineous marriage, presented with delayed speech following congenital hearing loss. There was history of missed abortion in mother and his first cousin had sensorineural hearing loss (SNHL) and hypopigmented patch over the left shoulder. Other milestones were normal. There was no history of limb defects, constipation, persistent vomiting or convulsions.

Physical examination revealed a white forelock (5 x 3.5 cm) on the central aspect of the forehead with a solitary café-au-lait spot (2 cm) over upper deltoid. The child also had mild synphrys, medial eyebrow hypertrichosis, white eyelash (polikilosis), hypertelorism, lateral displacement of lacrimal apparatus, heterochromia iridis (bluish) in both the eyes and dystopia canthorum (Figure 1). Waardenburg Syndrome index was 2.15 (significant).

![Figure 1](image-url)
Waardenburg Syndrome Type I: Case Report

W = X + Y + a/b = 2.15
X = (2a - 0.2119c - 3.909)/c = 0.4426
Y = (2a - 0.2479b - 3.909)/b = 0.823

(a = inner canthal distance = 4.9 cm; b = interpupillary distance = 5.5 cm; c = outer canthal distance = 9 cm)

Waardenburg Syndrome index greater than 1.95 is a major criteria and hallmark of Waardenburg Syndrome I. Child also had low set ears, left head slant with facial hemiatrophy, right head tilt, high medial root and long curling eyelashes (Figure 2). BERA revealed bilateral SNHL.

II. Discussion

Waardenburg Syndrome was founded in the Netherlands in 1951 when Petrus Johannes Waardenburg discovered that school children with different eye colour also had sensorineural hearing loss. It accounts for 2-3% of the congenital hearing loss. Other associated congenital anomalies are neural tube defects, Hirschprung’s disease, cleft palate, Sprengel shoulder, contractures, limb hypoplasia, urinary abnormalities and ptosis of the eyes. In 1992, the Waardenburg Syndrome Consortium proposed a diagnostic criteria for Waardenburg Syndrome I. Individuals are considered to have Waardenburg Syndrome Type I if they have 2 major or 1 major plus 2 minor criteria (2).

Major criteria:
• Born deaf or hard of hearing (congenital sensorineural hearing loss) (58% of individuals)
• Brilliant sapphire blue eyes (27%) or two different colour eyes (36%)
• White lock of hair on the forehead (17-58%)
• Immediate family member with Waardenburg Syndrome (37%)
• Inner corner of the eye displaced laterally (dystopia canthorum)

Minor criteria:
• Patches of light or white skin
• Eyebrows extending toward middle of the face (12%)
• Nose abnormalities (58%)
• Premature greying of the hair (by age 30)

Waardenburg Syndrome I is an autosomal dominant syndrome with abnormalities in PAX 3 gene.

III. Conclusion

This was a classical case of Waardenburg Syndrome Type I with five major criteria viz affected first degree relative, bilateral Sensory Neural Hearing Loss, heterochromia iridis, white forelock, and dystopia canthorum along with 2 minor criteria i.e synophrys and high nasal root.

References
[4]. Waardenburg Syndrome syndrome type 1 ,Ali Karaman MD1, Cihangir Aliagaoglu, MD2, Dermatology Online Journal 12 (3): 21

DOI: 10.9790/0853-14724546 www.iosrjournals.org 2 | Page